

Unified Medical Language System[®] (UMLS[®]) Basics

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U.S. Department of Health & Human Services



Schedule

8:30 - 9:30	Welcome and Introduction
9:30 - 10:00	SPECIALIST Lexicon & Lexical Tools
10:00 -10:30	Semantic Network
10:30 – 10:45	BREAK
10:45 - 11:30	NLM, the UMLS, and Health Data Standards
11:30 -12:00	Metathesaurus
12:00 - 1:00	LUNCH
1:00 -1:30	Metathesaurus, continued
1:30 - 2:45	MetamorphoSys & UMLS Tools
2:45 - 3:00	BREAK
3:15 - 4:45	MetamorphoSys & UMLS Tools Hands On
4:45 - 5:00	Questions, Help, and Evaluation

Acknowledgements

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UMLS Video

<http://www.nlm.nih.gov/research/umls/UMLS300kbps.swf>

The UMLS consists of

Metathesaurus

1 million+
biomedical
concepts
from over 100
sources

Semantic Network

135 broad
categories and
54 **relationships**
between
categories

SPECIALIST Lexicon & Tools

lexical
information and
programs for
**language
processing**

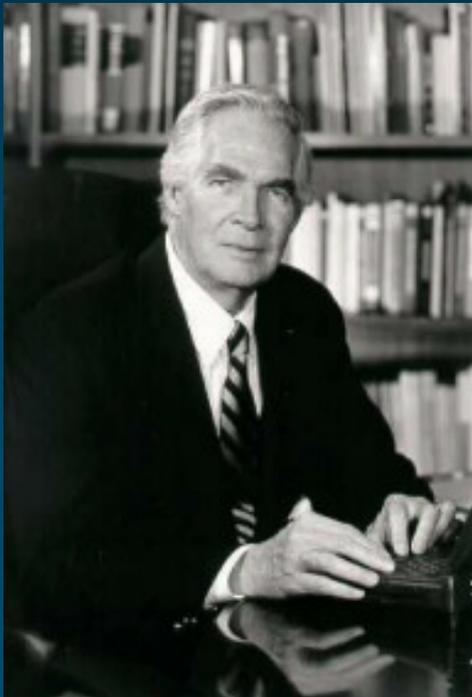
3 Knowledge Sources
used separately or together

History of the UMLS

[Lindberg & al., *Methods*, 1993]
[Humphreys & al., *JAMIA*, 1998]

- Started at National Library of Medicine, 1986
- “Long-term R&D project”
- Complementary to IAIMS

(Integrated Academic Information Management Systems)



«[...] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

- The first is the variety of ways the same concepts are expressed in different machine-readable sources and by different people.
- The second is the distribution of useful information among many disparate databases and systems.»

UMLS Objectives

- Intellectual “middleware”
- A set of multi-purpose tools for system developers
- Knowledge Sources used to overcome:
 - disparities in language format
Ex: atrial fibrillation, auricular fibrillation, af
 - disparities in granularity and perspective
Ex: Contusions, hematoma, bruise
Ex: Instruct patient to promptly report nosebleeds and excessive bruising (NIC), Epistaxis (MeSH)

UMLS Made available as:

- Data files
 - 3 separate sets of relational files
- Tools
 - MetamorphoSys (installation and customization)
 - RRF Subset Browser
 - lvg (lexical programs)
- Distributed on DVD; downloaded from UMLSKS

The UMLS is *not* an end-user application

UMLS Access

- UMLS Knowledge Source Server
 - <http://umlsks.nlm.nih.gov/>
 - Browser, Navigators
 - APIs
 - Download data files and programs
 - Documentation and other resources
- Local storage and customization (Metathesaurus)
 - MetamorphoSys
 - RRF Browser

UMLS Uses

- Information retrieval
- Thesaurus construction
- Natural language processing
- Automated indexing
- Electronic health records (EHR)
- Distribution mechanism for HIPAA, CHI, PHIN regulatory standards

2007AB UMLS: July 2007

- Metathesaurus:
 - 123 sources
 - 1,436,586 concepts
 - 17 languages
- Semantic Network:
 - 135 Semantic Types
 - 54 Semantic Relationships
- SPECIALIST Lexicon:
 - Over 297K records (over 482K inflectional forms)

A guided tour of UMLS Resources

- <http://www.nlm.nih.gov/research/umls/>
- <http://www.nlm.nih.gov/research/umls/documentation.html>
- http://www.nlm.nih.gov/research/umls/listserv_info.html
- <https://list.nih.gov/archives/umlsusers-l.html>
- http://www.nlm.nih.gov/research/umls/new_users.html

What is the UMLS?

Overview through an example

UMLS 3 Knowledge Sources

- Metathesaurus
 - Source vocabularies
 - Concepts
 - Relationships, Attributes
- Semantic Network
 - Semantic Types (categories)
 - Semantic Relationships
- Lexical resources
 - SPECIALIST Lexicon
 - Lexical tools

Metathesaurus: clusters terms by meaning

- Synonymous terms clustered into a concept
- Preferred term is chosen
- Unique identifier (CUI) is assigned

Addison's disease	Metathesaurus	PN	
Addison's disease	SNOMED CT	PT	363732003
Addison's Disease	MedlinePlus	PT	T1233
Addison Disease	MeSH	PT	D000224
Bronzed disease	SNOMED Intl 1998	SY	DB-70620
Deficiency; corticorenal, primary	ICPC2-ICD10 Thesaurus	PT	MTHU021575
Primary Adrenal Insufficiency	MeSH	EN	D000224
Primary hypoadreanlism syndrome, Addison	MedDRA	LT	10036696

C0001403

Addison's disease

Cluster of synonymous terms

Concept
C0001621

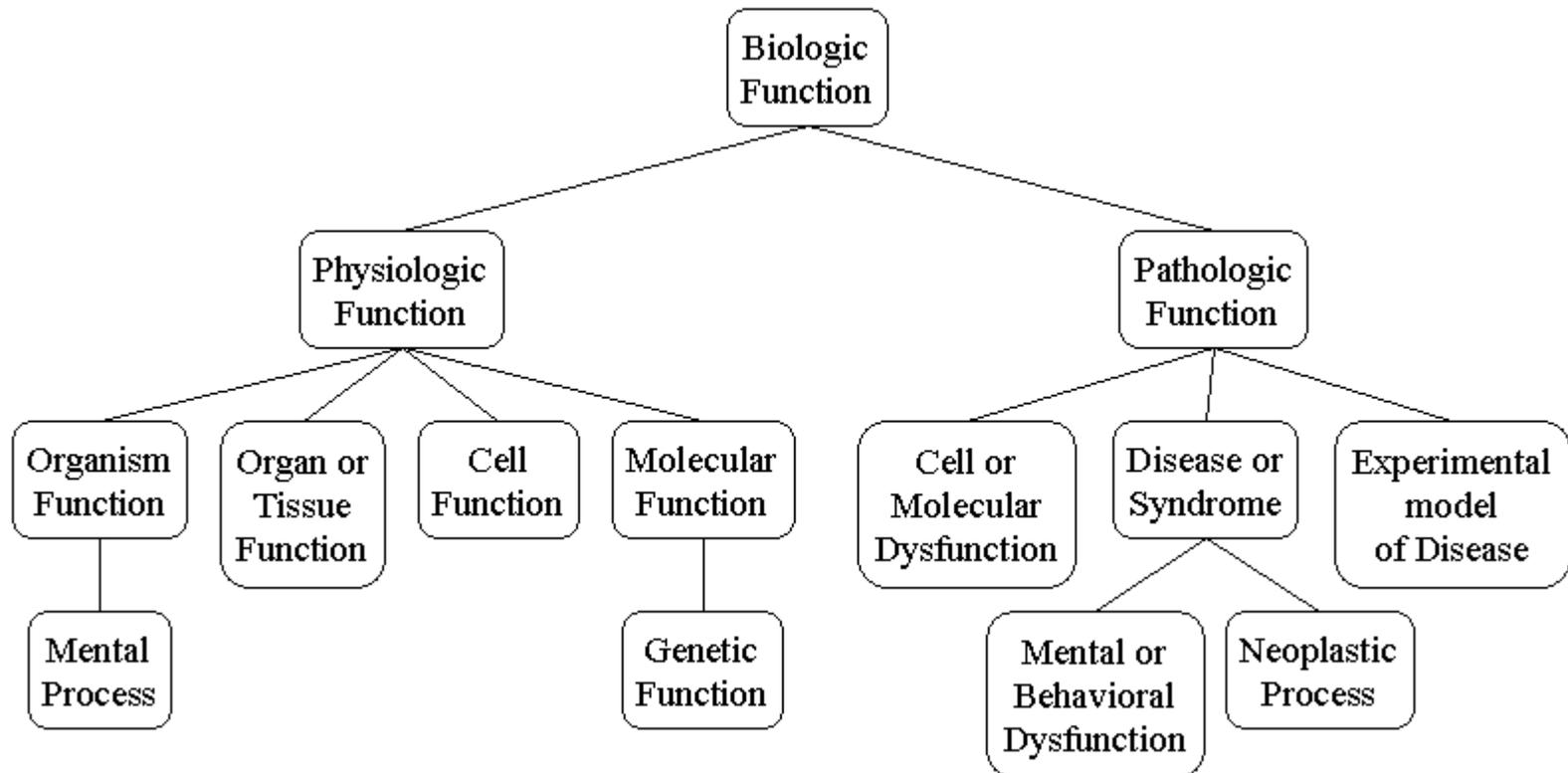
Term L0001621	<p>S0011232 <i>Adrenal Gland Diseases</i></p> <p>S0011231 Adrenal Gland Disease</p> <p>S0000441 Disease of adrenal gland</p> <p>S0481705 Disease of adrenal gland, NOS</p> <p>S0220090 Disease, adrenal gland</p> <p>S0044801 Gland Disease, Adrenal</p>	[...]
Term L0041793	<p>S0860744 <i>Disorder of adrenal gland, unspecified</i></p> <p>S0217833 Unspecified disorder of adrenal glands</p>	
Term L0161347	<p>S0225481 <i>ADRENAL DISORDER</i></p> <p>S0627685 DISORDER ADRENAL (NOS)</p>	[...]
Term L0181041	<p>S0632950 <i>Disorder of adrenal gland</i></p> <p>S0354509 Adrenal Gland Disorders</p>	[...]
Term L0368399	<p>S0586222 <i>Adrenal disease</i></p> <p>S0466921 ADRENAL DISEASE, NOS</p>	[...]
Term L1279026	<p>S1520972 <i>Nebennierenkrankheiten</i></p>	GER
Term L0162317	<p>S0226798 <i>SURRENALE, MALADIES</i></p>	FRE

[...]

Semantic Network

- 135 Semantic Types
 - Broad subject categories (Clinical Drug, Virus)
 - Ex:
 - Addison's Disease
 - Semantic Type: Disease or Syndrome
- 54 Semantic Relationships
 - Links between categories (isa, causes, treats)
 - Ex:
 - Virus causes Disease or Syndrome
- Types + Relationships
 - Form the structure of the semantic network
 - Broadly categorize the biomedical domain

Semantic Network: Biologic Function



SPECIALIST Lexicon

- General English lexicon of common words, biomedical terms
- Used by SPECIALIST Natural Language Processing System
- Lexical records contain encoded information:
 - syntactic (how words are put together),
 - morphological (inflection, derivation, and compounding), and
 - orthographic (spelling) information
- Lexical tools process terms
- Used in Metathesaurus production

Adrenal gland diseases
Adrenal disorder
Disorder of adrenal gland
Diseases of the adrenal
glands
C0001621

SPECIALIST Lexicon

```
{base=disease
  entry=E0023270
  cat=noun
  variants=req
  variants=uncount
  compl=pphr(of,np|bone|)
  compl=pphr(of,np|breast|)
  compl=pphr(of,np|liver|)
  compl=pphr(of,np|ovary|)
}
```

Summary

Metathesaurus:

- clusters terms into concepts – assigns unique identifier

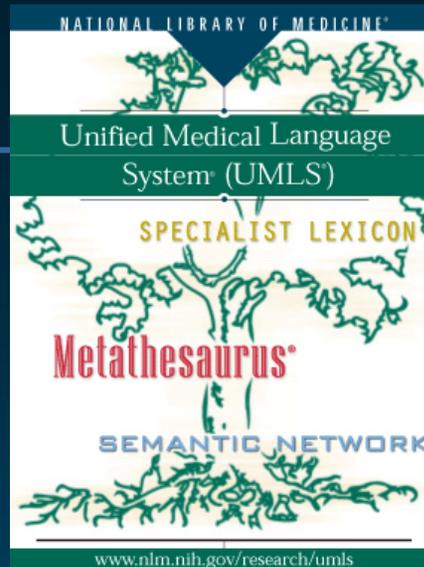
Semantic Network

- defines relationships between concepts, organizes concepts into categories

Lexicon and Lexical Tools:

- processes terms for entry into the Metathesaurus

UMLS Bookmark



The UMLS is a collection of tools and resources that enable **systems** to link biomedical information from different sources, such as patient record systems, bibliographic databases, factual databases, decision support systems and other biomedical and health applications.

The UMLS also improves the ability of computer programs to understand biomedical meaning in user inquiries, and to use that understanding to retrieve and integrate relevant information for users.

The UMLS consists of data files and programs organized into three Knowledge Sources. Those sources are:

METATHESAURUS — Contains information about biomedical concepts and terms from over 100 controlled vocabularies and classification, including U.S. clinical and administrative standards. Primarily in English, with some foreign language translations. Accompanied by MetamorphoSys, a program that can be used to produce customized versions for particular applications.

SEMANTIC NETWORK — Defines broad subject categories to which Metathesaurus concepts are assigned, as well as the useful relationships that exist between categories.

SPECIALIST LEXICON — Includes commonly occurring English and biomedical words, as well as information necessary for natural language processing, such as word form, structure, and spelling variants. Accompanied by a suite of lexical programs.

Together these Knowledge Sources and programs provide useful tools to overcome differences in biomedical terminologies that often create barriers to effective retrieval and integration of biomedical information.

The UMLS Knowledge Source Server (umlsks.nlm.nih.gov) provides Web access to the component parts of the UMLS as well as the ability to download UMLS files and programs for local use.

The UMLS does not include end-user applications beyond the Knowledge Source Server. Using the UMLS requires appropriate computing resources and technical expertise.

License Agreement — The UMLS is free of charge. Users complete a license agreement that protects the rights of the providers of component vocabularies. A separate license agreement is necessary with some sources.

ADDITIONAL INFORMATION

— umlsinfo.nlm.nih.gov

- Fact Sheets
- Online Documentation
- License Agreement
- Knowledge Source Server
- Other UMLS Information, e.g., FAQs, Learning Resources, UMLS Users Listserv

— custserv@nlm.nih.gov — NLM's e-mail address for questions on the UMLS and all other NLM resources



U.S. DEPARTMENT OF HEALTH
AND HUMAN SERVICES

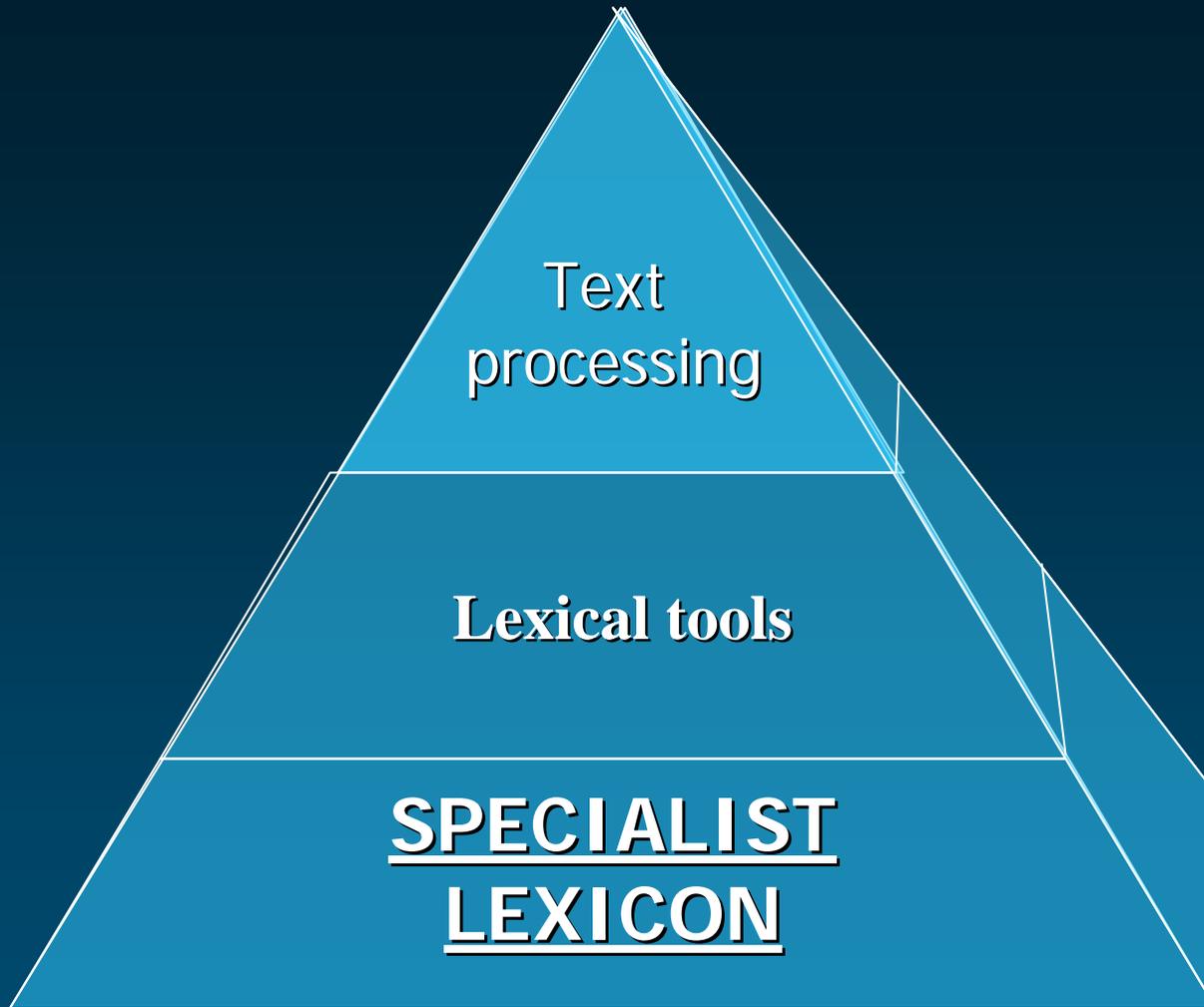
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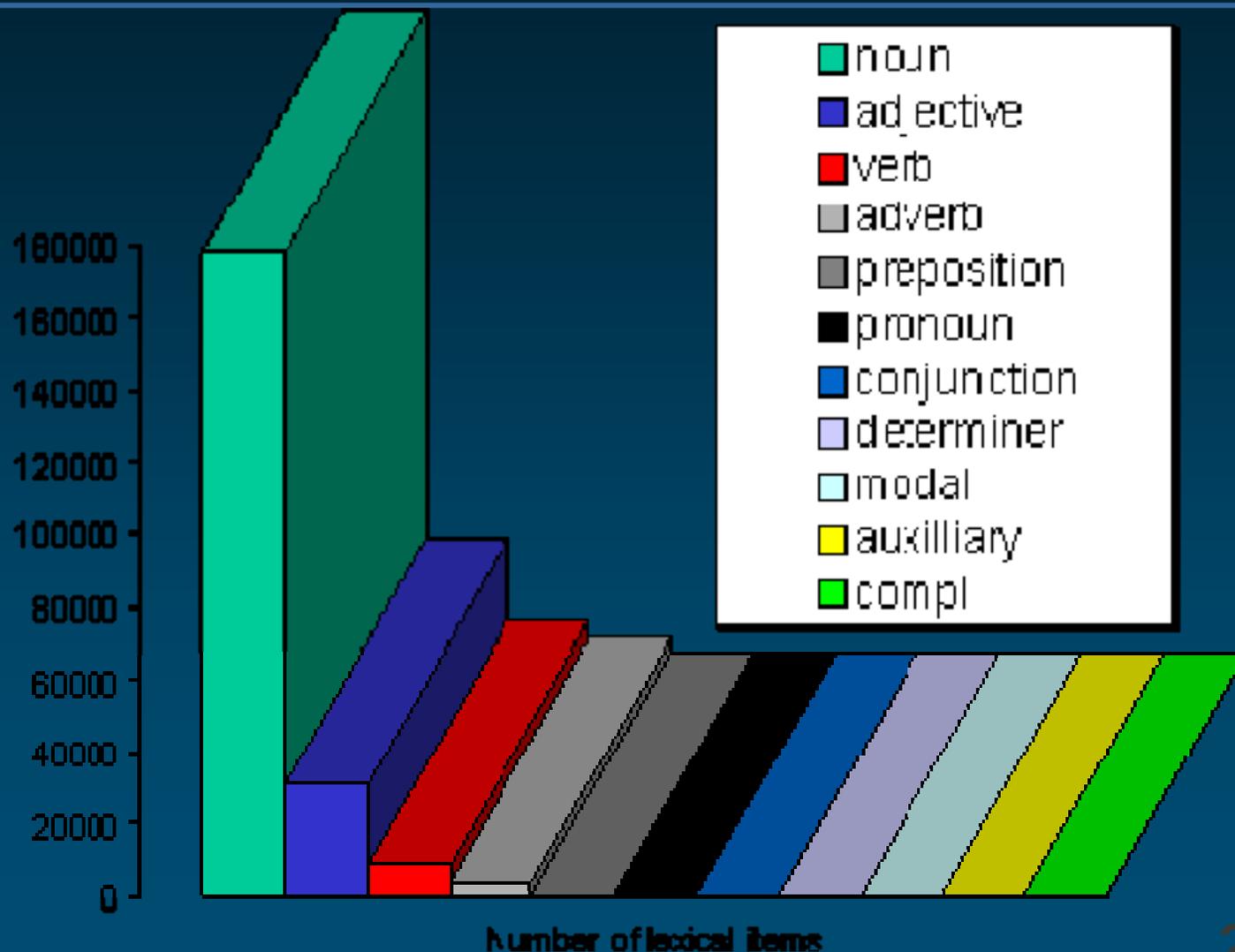
SPECIALIST Lexicon and Lexical Tools



SPECIALIST Lexicon

- Syntactic lexicon
 - 250K+ common and biomedical words (English) → 400K+ variants
 - Word properties
 - Syntax (how words are put together)
 - Morphology (inflection, derivation, and compounding)
 - Orthography (spelling)
- Used by SPECIALIST Natural Language Processing System to process text, terms
- Used in Metathesaurus production

SPECIALIST Lexicon by part of speech



Syntax

- Complementation

- verbs

- intransitive

I'll treat.

- transitive

He treated the patient.

- ditransitive

He treated the patient with a drug.

- nouns

- prepositional phrase: Valve of coronary sinus

- position for adjectives

Orthography

- Spelling variants

- oe/e

oesophagus - esophagus

- ae/e

anaemia - anemia

- ise/ize

cauterise - cauterize

- genitive mark

Addison's disease

Addison disease

Addisons disease

- British-American variants

criticise -- criticize

centre -- center

foetus -- fetus



SPECIALIST Lexicon

lexical records

```
{base=Kaposi's sarcoma  
spelling_variant=Kaposi sarcoma  
entry=E0003576  
  cat=noun  
  variants=uncount  
  variants=reg  
  variants=glreg  
}
```

```
{base=chronic  
entry=E0016869  
  cat=adj  
  variants=inv  
  position=attrib(1)  
  position=pred  
  stative  
}
```

```
{base=aspirate  
entry=E0010803  
  cat=verb  
  variants=reg  
  tran=np  
  nominalization=aspiration|noun|E0010804  
}
```

```
{base=in  
entry=E0033870  
  cat=prep  
}
```

Ancillary Data Bases and Rules Files

- Synonymy (sm.db)
genicular|adj|knee|noun
hepatocellular|adj|liver
cells|noun
- Derivation (dm.db,
dm.rules)
- Inflection (im.rules)
- Neoclassical compounds
(nc.db)

singular ends with:	plural ends with:	Examples
-us	-i	focus/foci
-ma	-mata	trauma/traumata
-a	-ae	larva/larvae
-um	-a	ilium/ilia
-on	-a	taxon/taxa
-sis	-ses	analysis/analyses
-is	-ides	cystis/cystides
-men	-mina	foramen/foramina
-ex	-ices	index/indices
-x	-ces	matrix/matrices



About

- [Home](#)
- [Lexical Systems Group](#)
- [Staff Directory](#)

Projects

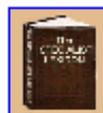
- [SPECIALIST Lexicon](#)
- [Lexical Tools](#)
- [Text Tools](#)
- [GSpell](#)

Documents

- [Papers & Reports](#)
- [Presentations](#)
- [Terms and Conditions](#)

The SPECIALIST NLP Tools

The SPECIALIST Natural Language Processing (NLP) Tools have been developed by the [The Lexical Systems Group](#) of the [National Center for Biomedical Communications](#) to investigate the contributions that natural language processing techniques make to the task of mediating between the language of users and the language of online biomedical information resources. The SPECIALIST NLP Tools facilitate natural language processing by helping application developers with lexical variation and text analysis tasks in the biomedical domain. The NLP Tools are open source resources distributed subject to these [terms and conditions](#).



A large syntactic lexicon of biomedical and general English, designed to provide the information needed for the SPECIALIST NLP System.



A set of JAVA programs designed to help users manage lexical variation, indexing, and normalization, etc. in biomedical text.



A nested set of JAVA objects designed to help users analyze free text documents into words, terms, phrases, and sections.



Includes two programs GSpell a spelling suggestion tool and BagOwordsPlus a phrase retrieval tool.

<http://lexsrv3.nlm.nih.gov/SPECIALIST/index.htm>

Lexical Tools

- Wordind
 - breaks strings into words
 - produces the Metathesaurus word indexes (MRXW)
- lvg
 - performs various lexical transformations
 - 58 flow components and 32 options in 2005
- NORM
 - a selection of LVG transformations
 - used for Metathesaurus indexing
 - produces Metathesaurus normalized word and string indexes (MRXNW & MRXNS)
 - used to access those indexes

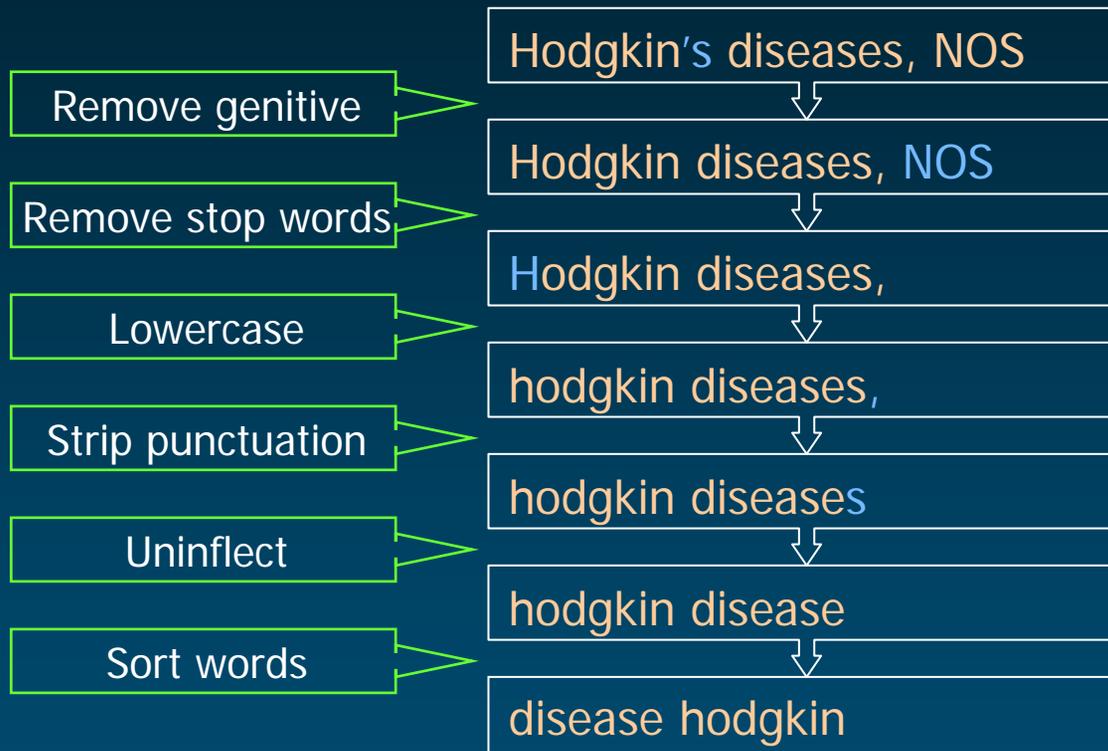
Lexical tools: wordInd

- Thyroxine unmasks Addison's disease
 - Thyroxine
 - unmasks
 - Addison
 - s
 - disease

C0001403|ENG|P|L0001403|PF|S0354372|N|A0388282||||SNM|SY|D-2332|Addison's disease|4|256|

- C0001403|S0354372|Addison's|
- C0001403|S0354372|disease|

Normalization



Normalization: Example

Hodgkin Disease
HODGKINS DISEASE
Hodgkin's Disease
Disease, Hodgkin's
Hodgkin's, disease
HODGKIN'S DISEASE
Hodgkin's disease
Hodgkins Disease
Hodgkin's disease NOS
Hodgkin's disease, NOS
Disease, Hodgkins
Diseases, Hodgkins
Hodgkins Diseases
Hodgkins disease
hodgkin's disease
Disease, Hodgkin

normalize

disease hodgkin

Normalized
term is not
necessarily
readable

Spelling Suggestion Tools

- **GSpell**
 - term retrieval tool
 - common misspellings
- **BagOWordsPlus**
 - phrase retrieval tool
 - uses correctly spelled words within the phrase to limit possible candidates
 - uses GSpell when it has to

Lexical Systems Group

Official Websites	http://SPECIALIST.nlm.nih.gov http://mmtx.nlm.nih.gov
Mail Addresses	Allen Browne - browne@nlm.nih.gov Guy Divita - divita@nlm.nih.gov Chris Lu - lu@nlm.nih.gov Lexical Systems group - umlslex@nlm.nih.gov

Semantic Network

Semantic Network

- **Semantic Types**

- 135 broad subject categories (Clinical Drug, Virus)
- Organized into 2 hierarchies: entity, event
 - Addison's Disease (concept)
 - Semantic Type: Disease or Syndrome

- **Semantic Relationships**

- 54 possible, useful links between categories
- Hierarchical: isa
- Associative, e.g. causes
 - Virus **causes** Disease or Syndrome

- **Types + Relationships = Semantic Network**

- <http://www.nlm.nih.gov/research/umls/meta3.html>

Semantic Types categorize

- Concept: Discipline of Nursing
- Semantic Type: **Biomedical Occupation or Discipline**

- Concept: Nursing Homes
- Semantic Type: **Health Care Related Organization**
Manufactured Object

- Concept: Home Nursing
- Semantic Type: **Health Care Activity**

Semantic Type: Disease or Syndrome

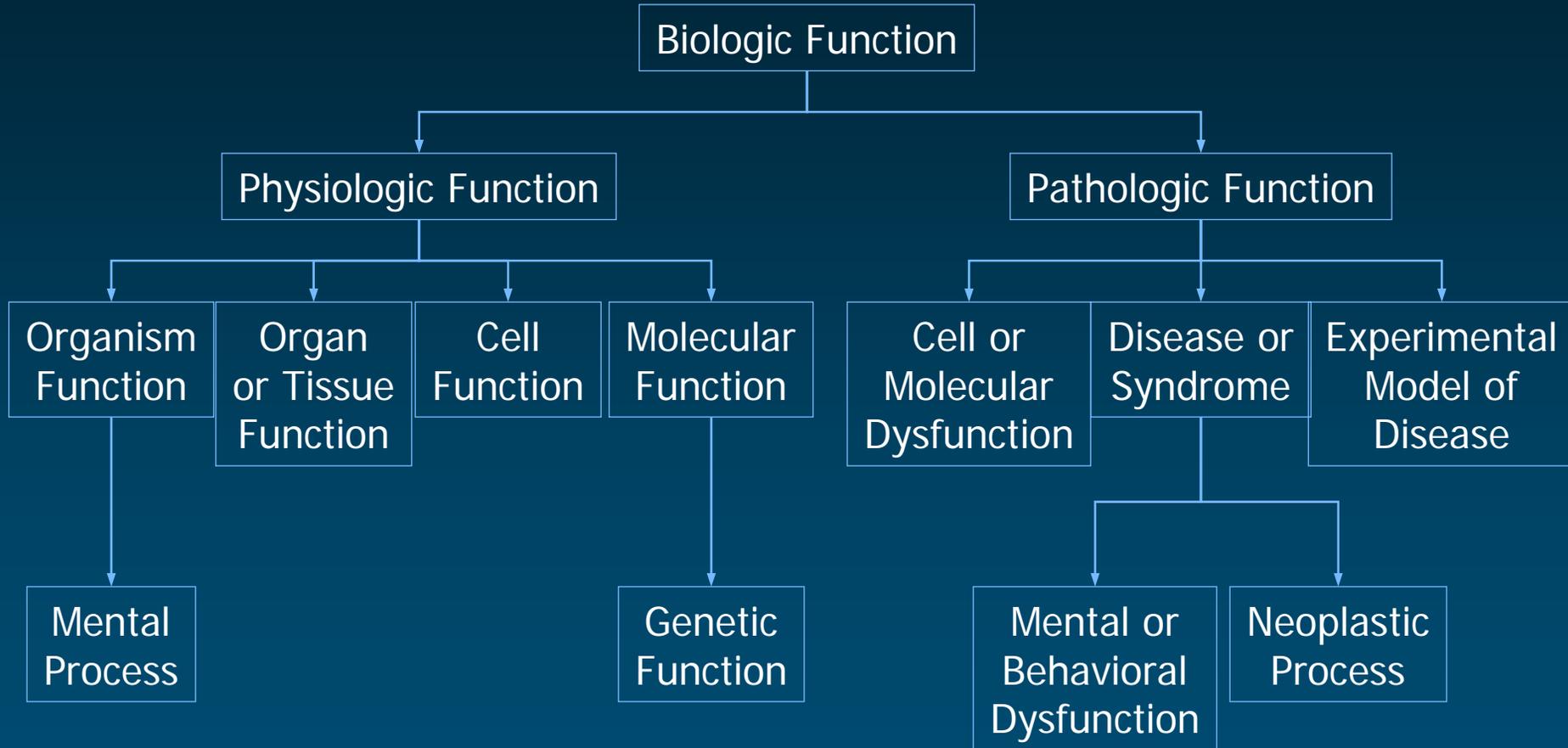
- Natural Phenomenon or Process
 - Biologic Function
 - Physiologic Function
 - Organism Function
 - » Mental Process
 - Organ or Tissue Function
 - Cell Function
 - Molecular Function
 - » Genetic Function
 - Pathologic Function
 - *Disease or Syndrome* ←
 - Mental or Behavioral Dysfunction
 - Neoplastic Process

54 Semantic Relationships

[link](#)

- **Hierarchical** (isa = is a kind of)
 - among types
 - Animal *isa* Organism
 - Enzyme *isa* Biologically Active Substance
 - among relations
 - treats *isa* affects
- **Non-hierarchical**
 - Sign or Symptom *diagnoses* Pathologic Function
 - Pharmacologic Substance *treats* Pathologic Function

“Biologic Function” hierarchy (isa)



Semantic Relationships

Define useful relations between types

Biomedical Occupation or Discipline

ASSOCIATED_WITH Social Behavior

Language

ISSUE_IN

Biomedical Occupation or
Discipline

Manufactured Object

CAUSES

Injury or Poisoning

Diagnostic Procedure

USES

Manufactured Object

Health Care Activity

ISA

Occupational Activity

Health Care Activity

AFFECTS

Disease or Syndrome

Semantic Relationships

Disease or Syndrome associated with Finding

Disease or Syndrome result of Pathologic Function

Body Part, Organ, or Organ Component location of
Disease or Syndrome

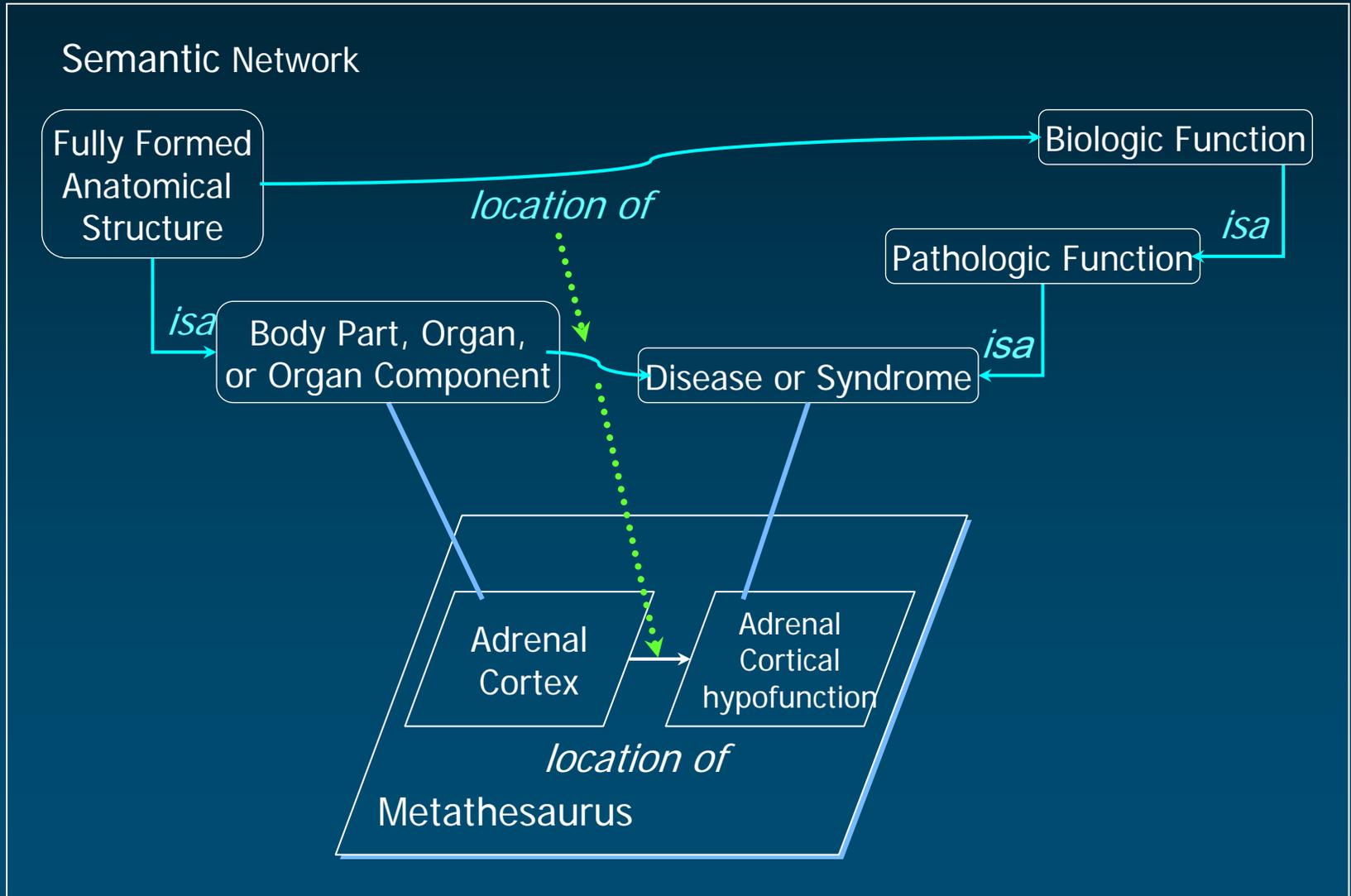
Hormone affects Disease or Syndrome

Hormone complicates Disease or Syndrome

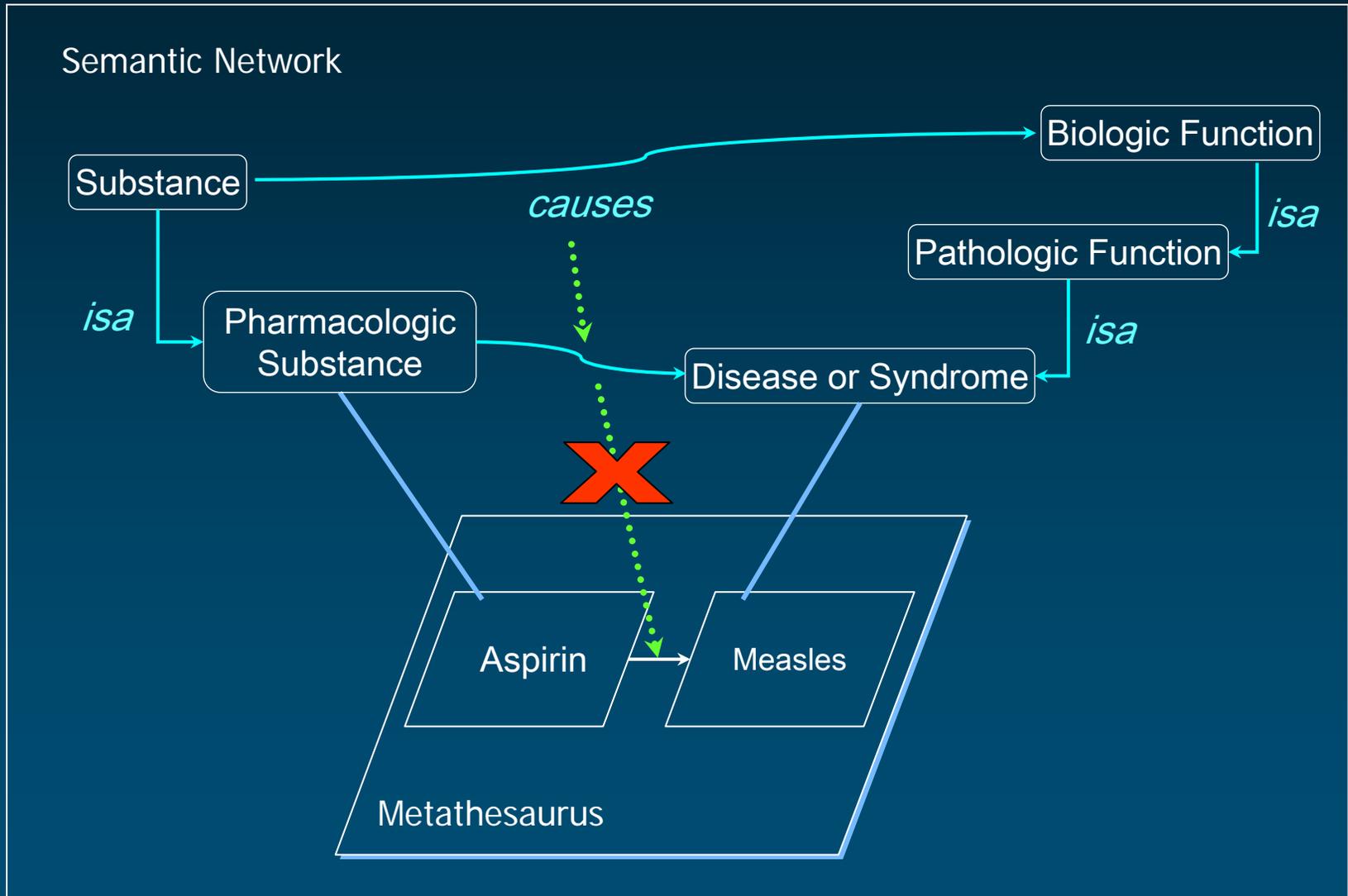
Laboratory Procedure diagnoses Disease or Syndrome

Pharmacologic Substance treats Disease or Syndrome

Relationships can inherit semantics



Relationships don't always inherit semantics



Why have a Semantic Network?

- Semantic Type serves as a **high level category** assigned to each Metathesaurus concept, *independent of its position in its original source hierarchy*
- A Semantic Relation is a **possible link between 2 concepts** assigned those Semantic Types
 - The relationship may hold at the concept level
 - Other relationships may apply at the concept level

Semantic Network Data Files

ASCII relational files

- SRDEF: Basic information about Semantic Types and Relations
- SRSTR: Structure of the Network
- SRSTRE1: Fully inherited set of Relations (UI's)
- SRSTRE2: Fully inherited set of Relations (names)
- SRFIL: Description of each table
- SRFLD: Description of each field and table(s) in which it is found

Unit records

- SU Semantic Types, Relationships

UMLS and Health Data Standards

The call for standards

- 1991 – IOM: *The Computer-Based Patient Record*
- 1995 – PHS: *Making a Powerful Connection*
- 1996 – Congress: *HIPAA*
- 2000 – IOM: *To Err is Human*
- 2000 – NCVHS: *Report on Standards for PMRI*
- 2001 – IOM: *Crossing the Quality Chasm*
- 2002 – NCVHS: *PMRI recommendations (1st Set)*
- 2002 – AMIA Congress: *Drug Nomenclature Information*
- 2002 – AAMC conference: *IT Enabling Clinical Research*
- 2003 – CHI eGov: *U.S. gov't target standards (1st set)*
- 2003 – Connecting for Health: *Standards Wk.Gr.Report*

Strategy for U.S. health data standards

- Establish mechanisms for designating U.S. standards
 - ☐ ✓ Pick best available as starting point
 - ☐ ✓ Support development, maintenance, distribution
- Coordinate development of selected standards to achieve non-overlapping, interlocking set
- Broaden participation in standards development
- Promote use and improvement

Federal standards selection mechanisms

- **HIPAA** (1996) - Health Insurance Portability and Accountability Act of 1996 requires administrative standards
- **NCVHS** - National Committee on Vital and Health Statistics, a long-standing (50+ years) advisory committee to HHS expanded by HIPAA, recommends standards
- **CHI** (2001) Consolidated Health Informatics project, a cross-agency eGov initiative designates U.S. gov't-wide clinical data standards
- **Medicare Modernization Act** (2003) - requires e-Prescribing standards; establishes Commission on Systemic Interoperability

Standards implementation

- **CHI (clinical)** *LOINC*
 - E.g., lab test results, problems, diagnoses, history, physical
 - Electronic exchange of clinical health information in U.S. Government systems
- **HIPAA (administrative)** *CPT*
 - e.g., health insurance claims, billing, ordering
 - HIPAA Administrative Simplification provisions
 - Designated DHHS national standards for electronic healthcare transactions
- **PHIN (public health)** *ICD-9-CM*
 - e.g., disease surveillance, immunization rates, environmental monitoring
 - CDC designated standards for public health reporting

Medical terminology standards

- Key element in electronic medical records systems
- 2003 agreement between NLM and College of American Pathologists for U.S. rights to SNOMED CT
- 2007 International Health Terminology Data Standards Organisation purchases SNOMED CT

NLM role

- HHS central coordinating body for Patient Medical Record Information (PMRI) terminologies
- Work with agencies, organizations to support standards
- Support mappings between standard clinical vocabularies (SNOMED, LOINC, RxNorm) and others
- Reduce overlap between standards vocabularies
- Maintain UMLS as uniform distribution mechanism for
 - HIPAA code sets, recommended PMRI terminologies
 - **mappings** between standard codes and PMRI terminologies
 - information on valid vocabularies for transaction standards

SNOMED CT in the UMLS

- English descriptions and relationships (2004)
 - Concepts: 298,090
 - Descriptions: 736,946 (synonyms)
 - Relationships: 1,315,910
- Added to Metathesaurus:
 - Concepts: 37,089
 - Descriptions: 350,464
- Spanish terms added in 2004AB

SNOMED CT in the Metathesaurus

- Core content U.S. English, Spanish
 - Concepts, Descriptions, Relationships tables
 - Generic drugs
 - History table
 - ICD-9-CM mapping
- SNOMED updates in UMLS
 - January SNOMED CT update → Spring UMLS
 - July SNOMED CT update → Fall UMLS

Additional information on NLM Web site

- [SNOMED CT[®] in the UMLS[®] Metathesaurus[®]: Inversion Source Transparency Achieved Jan. 2005](#)
- [SNOMED CT in the 2005AB Release](#)
- [SNOMED CT[®] in the UMLS[®] Metathesaurus[®]:Release Source Transparency Achieved Jan. 2005](#)
- [SNOMED CT in the 2005AB Release](#)
- [SNOMED CT Information from the College of American Pathologists](#)

RxNorm

NLM's RxNorm Project

- Developed by NLM to address
 - missed synonymy in UMLS clinical drugs
 - medication errors
- CHI recommended standard
- Intended to support
 - effective sharing of drug data across systems
 - electronic health record (EHR)
 - computerized physician order entry (CPOE)

RxNorm

- RxNorm (standalone vocabulary)
 - Normalized forms of clinical drugs
 - Created by NLM
 - From commercial and government clinical drug vocabularies
- RxNorm subset (extracted from UMLS)
 - RxNorm forms (NLM)
 - Clinical drug content including SNOMED CT, VANDF, FDA NDC

Respect license restrictions

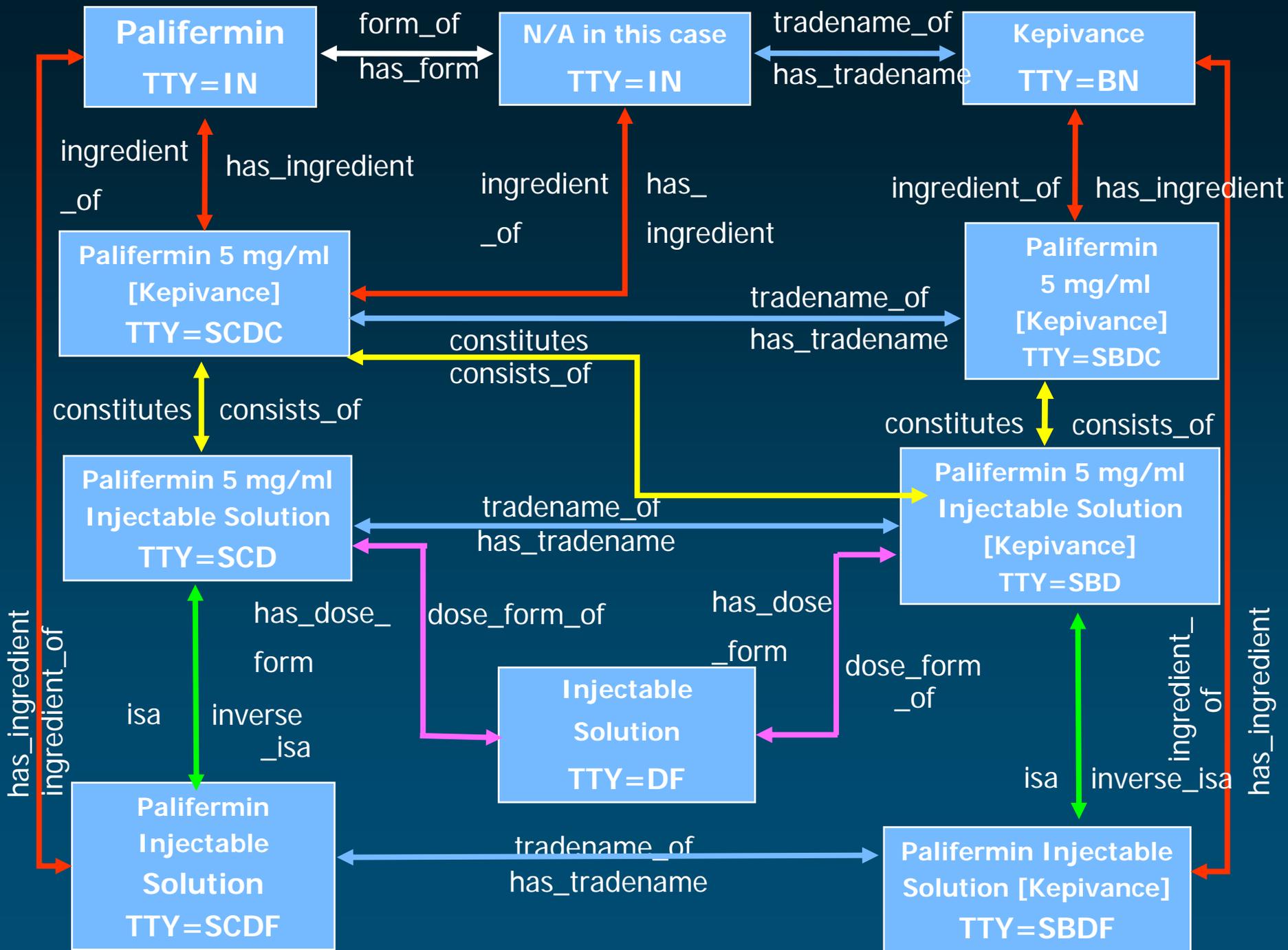
RxNorm Normalized Forms

- Semantic Normal Form (SNF)
 - Active standardized (generic) ingredients
 - Strengths
 - Units of measurement
 - Dosage form
- As administered to a patient
 - Acetaminophen 500 MG Oral Tablet
 - Acetaminophen 500 MG Oral Tablet [Tylenol]

RxNorm Term Types

- SBD = Palifermin 5 mg/ml Injectable Solution [Kepivance] → 7 additional concepts
- IN= Palifermin
- BN= Kepivance
- SBDC= Palifermin 5 mg/ml [Kepivance]
- SBDF = Palifermin Injectable Solution [Kepivance]
- SCD= Palifermin 5 mg/ml Injectable Solution
- SCDC= Palifermin 5 mg/ml
- SCDF= Palifermin Injectable Solution

all are separate RxNorm concepts



Search



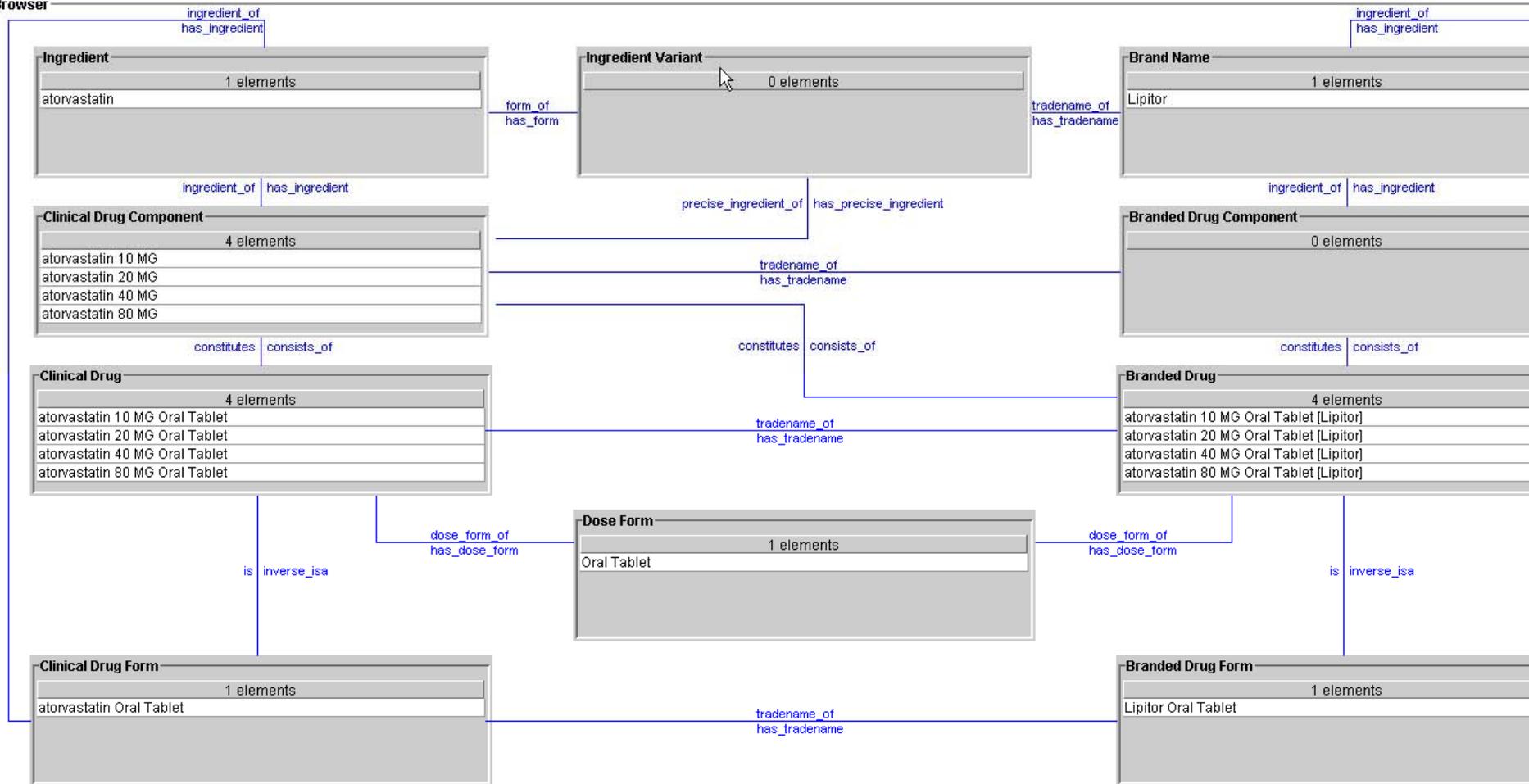
Please enter the search term:

Lipitor

Search



Browser



RxNorm (Source)

- Full sources
 - FDA, NDDF, Medispan, Micromedex, Multum
 - More to come
- Partial sources
 - SNOMED CT, MeSH
- NOT in RxNorm
 - USP Medicare Model Guidelines
 - MedDRA (adverse reactions)

RxNorm (NLM)
SNOMED CT (CAP)
MTHFDA (FDA NDC)
VANDF (VA)
Gold Standard
NDDF (FirstDataBank)
MDDB (Medispan)
MMX (Micromedex)
Multum

Mappings

Mappings

- Synonymy within concept (implied)
- Explicit links between concepts, terms, codes or other content in specified vocabularies
- Source-asserted
 - SNOMED CT → ICD-9-CM
- Created/supported by NLM
- Represented in
 - MRREL (simple only)
 - MRMAP, MRSMAP (simple and complex)

UMLS – MeSH mapping file

Used in MEDLINE/PubMed searching

Based on synonymy

...

myocardial infarction|attack coronary

myocardial infarction|attack heart (nos)

myocardial infarction|cardiac infarction

myocardial infarction|cardiac infarction, nos

myocardial infarction|cardiac; infarction

myocardial infarction|heart attack

myocardial infarction|heart attack, nos

myocardial infarction|heart attacks

myocardial infarction|heart infarction

Mapping projects planned/underway

- CHI standards → HIPAA code sets
 - SNOMED CT → ICD-9-CM (support reimbursement)
 - SNOMED CT → ICD-10-CM
 - SNOMED CT → CPT/HCPCS
 - LOINC → CPT

- SNOMED CT →
 - MedDRA
 - MeSH
 - Nursing (NIC, NOC, NANDA)

Mapping examples

LOINC - CPT

- 1795-4 AMYLASE:CCNC:PT:FLU:QN →
- 1798-8 AMYLASE:CCNC:PT:SER:QN →
- 1799-6 AMYLASE:CCNC:PT:UR:QN →
- 82150 Amylase (blood or urine)

SNOMED CT – CPT

- 313500004 Urine protein/creatinine ratio measurement →
- 82570 Creatinine; other source
- 84155 Protein, total, except by refractometry; serum

Key NLM Assumptions about Mappings

- Participants must include **producers** on both ends and **users**
- Mappings may prompt changes to content and adjustment to update schedules
- Mappings must keep current at both ends
- Mappings will be distributed in the UMLS with license terms applicable to both ends
- Mapping is an R & D problem; iteration is required to build highly functional maps

Toward interoperability

- Office of the National Coordinator of Health Information Technology (ONC)
- Standards harmonization
- Clear policies about data control and ownership
- Common security practices
- Security of vendor products
- Interoperability as a business



Versel, Neil. "Brailer Uses AMIA Meeting as Bully Pulpit for Change," Health-IT World News, Tuesday, April 12, 2005.

Electronic health records (EHR)

- National collaboration toward secure, Internet-accessible EHR in 10 years
- Federal and private-sector to develop standards with agreement from all stakeholders
- Electronic health records and other information technology to transform health care system
 - reduce medical errors, improve quality of care
 - minimize paperwork hassle, lower costs

Secretary Leavitt Takes News Steps to Advance Health IT, June 15, 2005 (www.hhs.gov/healthit)

Metathesaurus

Metathesaurus

- The Metathesaurus is a
 - very large
 - multi-purpose
 - multi-lingual biomedical vocabulary

- It contains information about
 - biomedical and health related concepts
 - their various names
 - the relationships among them

Metathesaurus Sources

- Over 100 controlled vocabularies, code sets, and mappings
- Determine scope of Metathesaurus
- Source meanings, hierarchies preserved
- Relationships, Semantic Types added

Metathesaurus Sources 2

- 119 vocabularies in 17 languages
- Broad coverage of general, specialized biomedicine
- Varying purposes, structures, properties
- Sets of valid values:
 - Thesauri, e.g., MeSH, CRISP, NCI
 - Statistical classifications, e.g., ICD-9-CM
 - Billing codes, e.g., CPT, ABC Codes
 - Clinical coding systems, e.g., SNOMED CT
- Complete source lists in Appendixes A.1, B.4

<http://www.nlm.nih.gov/research/umls/documentation.html>

Metathesaurus Sources 3

- Sources reflect regular and irregular update schedules
- NLM resources determine currency of sources in the Metathesaurus
- Priority given to maintaining critical standards vocabularies

Metathesaurus Sources 4

Why so Many?

- Unique applications
- Different types of information
- Levels of specificity
- Organizational and political boundaries

Why Not Combine Them Into One?

- One size does not fit all
- NLM supports coordination when possible
- Growing awareness of benefits of standardization

“The UMLS approach assumes continuing diversity in the formats and vocabularies of different information sources and in the language employed by different elements of the biomedical community. It is not an attempt to build a single standard biomedical vocabulary.”



Humphreys, BL and PL Schuyler, The Unified Medical Language System: Moving beyond the vocabulary of bibliographic retrieval. In: Broering NC, ed. High- Performance Medical Libraries: advanced information management for the virtual era. Westport (CT): Meckler; 1993, p. 33.

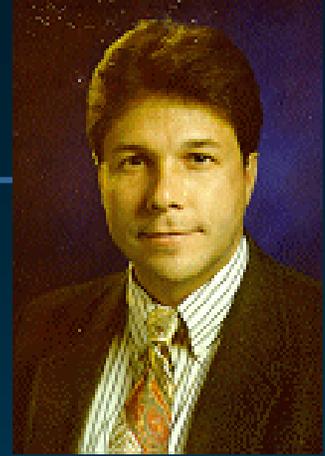
Metathesaurus Source Evaluation 1

1. Who does the vocabulary serve?
2. Is the vocabulary authoritative?
3. Is the thesaurus available in a well-structured, computable electronic form?
4. Have copyright ownership and license restriction levels been considered?

Metathesaurus Source Evaluation 2

5. What thesaurus characteristics and principles are used?
6. How fully descriptive are the terms when seen alone, rather than in the associated context?
7. Will there be many cases where the 'face' meaning of your terms are different from the meaning of the same term in other areas of biomedicine?
8. Who would be available to assist in editing and review?

Cimino's Desiderata



- Dr. James J. Cimino, M.D.
- <http://www.dbmi.columbia.edu/cimino/>
- Cimino JJ. Desiderata for Controlled Medical Vocabularies in the Twenty-First Century. *Methods Inform Med* 1998; 37: 394-403.
- Content
- Concept Orientation
- Concept Permanence
- Meaningless Concept Identifiers
- [.....]

Source data in Metathesaurus files

Names, Synonyms

Terms, Codes → MRCONSO

Relationships → MRREL

Hierarchies → MRHIER

Mappings → MRREL, MRMAP,
MRSMAP

Attributes → MRSAT

Definitions → MRDEF

CUIs links concept data across files

Metathesaurus files

- Concepts (MRCONSO)
 - Synonymous and near-synonymous terms
 - Codes
 - Concept Unique Identifier (CUI), Semantic Type added
- Relationships and mappings (MRREL, MRHIER, MRMAP, MRSMAP)
 - Concepts are related to other concepts
 - Source hierarchies are represented
 - Relationship attributes specify type, source of relationship
 - Simple and complex maps
- Attributes (MRSAT), (MRDEF)

Concept structure

Concept (1.3 M)
CUI
set of synonymous names

Term (> 4.6 M) LUI
set of normalized names

String (> 5.1 M) SUI
distinct concept name

Atom (> 6.2 M)
AUI
concept name in source

A1412439 headaches (BI)
S1459113 headaches

A2882187 Headache (SNOMED)
A0066000 Headache (MeSH)
S0046854 Headache

L0018681 headache

A1641293 Cranial Pain (MeSH)
S1680378 Cranial Pain

L1406212 cranial pain

A0418053
HEAD PAIN CEPHALGIA (DxP)
S0375902
HEAD PAIN CEPHALGIA

L0290366 cephalgia head pain

C0018681 Headache

Concept

cluster of synonymous terms

Concept
C0001621

Term
adrenal disease gland
L0001621

S0011232 *Adrenal Gland Diseases*
S0011231 Adrenal Gland Disease
S0000441 Disease of adrenal gland
S0481705 Disease of adrenal gland, NOS
S0220090 Disease, adrenal gland
S0044801 Gland Disease, Adrenal

Term
adrenal disorder gland
unspecified
L0041793

S0860744 *Disorder of adrenal gland, unspecified*
S0217833 Unspecified disorder of adrenal glands

Term
adrenal disorder
L0161347

S0225481 *ADRENAL DISORDER*
S0627685 DISORDER ADRENAL (NOS)

Term
adrenal disorder gland
L0181041

S0632950 *Disorder of adrenal gland*
S0354509 Adrenal Gland Disorders

Term
L0162317

S0226798 *SURRENALE, MALADIES* FRE

Metathesaurus metadata, history and indexes

- Metadata files
 - MRSAB sources
 - MRFILES files
 - MRCOLS data elements
 - MRDOC finite values of selected data
 - MRRANK source/term type precedence
- Change files deleted, merged CUI, LUI
- MRCUI.RRF retired CUIs
- Concept name indexes

Representing ambiguity multiple meanings

- Same string, different meanings
- Cold (L0009264)
 - cold temperature (C0009264)
 - common cold (C0009443)
 - chronic obstructive lung disease (C0024117)
 - Cold sensation (C0234192)
- AMBIGLUI.RRF (AMBIG.LUI)
 - L0009264|C0009443|
 - L0009264|C0024117|
 - L0009264|C0234192|
- AMBIGSUI.RRF (AMBIG.SUI)
 - S0026353|C0009443|
 - S0026353|C0234192|

Metathesaurus Distribution Formats

- Pipe-delimited text files
- UTF-8 character encoding

- Original Release Format (ORF) **or** Rich Release Format (RRF)

- Original Release Format (ORF)
 - **Metathesaurus-Concept-Centric view**
 - Explicit concept-based connection between terms in different sources
 - Most information represented at concept (CUI) level

Rich Release Format (RRF)

- **Source-Centric View** supports source transparency
- Represents original source information at atom (AUI) level
- Atom = unit of meaning (name) in a source
- New data fields and files to capture source specific identifiers, mappings

Metathesaurus highlights

- Concept based
- Represents the meaning in each source
- Represents data in common format
- Adheres to principle of “source transparency”

- Tags source information
- Adds context-free unique identifiers

- Includes normalized word and string indexes produced using UMLS lexical tools

Accessing the Metathesaurus

- UMLSKS <http://umlsks.nlm.nih.gov/>
 - Simple and advanced searches
 - View concept reports and raw relational records

- RRF Browser in MetamorphoSys
 - Generate and view customized subset
 - Search and browse
 - View concept reports and raw relational records

Metathesaurus License Agreement 1

- Online Web-based license:

<http://www.nlm.nih.gov/research/umls/license.html>

- Read license
- Read appendix 1
- Read appendix 2
- Print a copy for your records
- Complete the Web form

Accept and continue

Accept and continue

Accept

 **Printer-friendly Version**

Submit

- Verification and turnaround:

- Receive e-mail from NLM and respond within 72 hours
- NLM official countersigns, license added to database
- Receive 2nd e-mail from NLM with new license number

Metathesaurus License Agreement 2

- 2. No charges, usage fees or royalties will be paid to NLM.
- 5. Within 30 days of the end of any calendar year ... provide NLM with a brief report
- 11.c. required to include ... identifiers from ... the original source vocabularies
- 12. For material ... from some sources additional restrictions ... may apply.
- Special restrictions for SNOMED CT in light of the International Agreement

See list of current sources in Appendix A.1.

License Restriction Levels 0-4

- Level 0 (28.2%)

- *unrestricted*

- Level 1 (1.6%)

- *negotiate to translate*

- Level 2 (0.4%)

- *negotiate to use in health data creation*

- Level 3 (30.6%)

- *negotiate to use in production*

- *explicitly prohibited to provide Internet access*

- Level 4 (39.2%)

- *unrestricted for U.S. use and distribution*

There may be additional restrictions, or separate license fees, associated with usage of specific vocabularies. Read the UMLS License Agreement, including the Appendix!

67%

Metathesaurus Data Files

MRCONSO (sample rows 1..5)

(2004AB)

	1	2	3	4	5	6	7	8	9	10	11
	CUI	LAT	TS	LUI	STT	SUI	ISPRF	AUI	SAUI	SCUI	SDUI
1	C0001403	ENG	P	L0001403	PF	S0354372	Y	A4367951			
2	C0001403	ENG	P	L0001403	PF	S0354372	N	A2922421	485624014	363732003	
3	C0001403	ENG	P	L0001403	VC	S0010794	Y	A0019740		M0000346	D000224
4	C0001403	ENG	S	L0494851	PF	S2164152	N	A2018589			
5	C0001403	FRE	P	L3246333	PF	S3773545	Y	A3996251			D000224

	12	13	14	15	16	17	18
	SAB	TTY	CODE	STR	SRL	SUPPRESS	CVF
1	MTH	PN	NOCODE	Addison's disease	0	N	
2	SNOMEDCT	PT	363732003	Addison's disease	4	N	
3	MSH	MH	D000224	Addison's Disease	0	N	
4	MDR	LT	10052381	Primary adrenal insufficiency	3	N	
5	MSHFRE	MH	D000224	Addison, maladie	3	N	

MRCONSO (sample rows 6..10) (2004AB)

	1	2	3	4	5	6	7	8	9	10	11
	CUI	LAT	TS	LUI	STT	SUI	ISPRF	AUI	SAUI	SCUI	SDUI
6	C0001403	FRE	S	L1272481	PF	S1514427	Y	A1464383			
7	C0001403	GER	P	L1229627	PF	S1471573	Y	A4030156			D000224
8	C0001403	GER	S	L1239271	PF	S1481217	Y	A4034094			D000224
9	C0001403	JPN	P	L3437833	PF	S3965327	Y	A4264008			D000224
10	C0001403	JPN	S	L3465347	PF	S3992841	Y	A4291522			D000224

	12	13	14	15	16	17	18
	SAB	TTY	CODE	STR	SRL	SUPPRESS	CVF
6	WHOFRE	IT	0410	MALADIE D'ADDISON	2	N	
7	MSHGER	MH	D000224	Addison-Krankheit	3	N	
8	MSHGER	SY	D000224	Bronzehautkrankheit	3	N	
9	MSHJPN	MH	D000224	Addison病	3	N	
10	MSHPJN	SY	D000224	副腎性黒皮症	3	N	

MRCONSO (sample rows 11..13)

(2004AB)

	1	2	3	4	5	6	7	8	9	10	11
	CUI	LAT	TS	LUI	STT	SUI	ISPRF	AUI	SAUI	SCUI	SDUI
11	C0001403	POR	P	L3302998	PF	S3831123	N	A6382080			
12	C0001403	RUS	P	L3336992	PF	S3864473	Y	A4157629			
13	C0001403	SPA	P	L1226877	PF	S1468823	Y	A1419475			

	12	13	14	15	16	17	18
	SAB	TTY	CODE	STR	SRL	SUPPRESS	CVF
11	MDRPOR	LT	1001130	Doença de Addison	3	N	
12	MSHRUS	MH	D000224	АДДИСОНОВА БОЛЕЗНЬ	3	N	
13	WHOSPA	IT	0410	ADDISON, ENFERMEDAD	3	N	

MRHIER (sample rows)

(2004AB)

	1	2	3	4	5	6
	CUI	AUI	CXN	PAUI	SAB	RELA
1	C0001403	A0019740	1	A0020270	MSH	
2	C0001403	A0019740	2	A0028022	MSH	
3	C0001403	A0019743	3	A1988358	PSY	member_of_cluster
4	C0001403	A2922421	1	A3307650	SNOMEDCT	isa
5	C0001403	A2922421	2	A3307650	SNOMEDCT	isa

	7	8	9
	PTR	HCD	CVF
1	A0434168.A2367943.A2366890.A0135391.A0054194.A0020267.A0020270	C19.053.264.263	
2	A0434168.A2367943.A2366890.A0135391.A0072566.A0028022	C20.111.163	
3	A0449751.A1988279.A1988358		
4	A3684559.A3886745.A2880798.A3398606.A3399335.A3398961.A2872359. A2872360.A3307650		
5	A3684559.A3886745.A2880798.A3398606.A3399335.A3398961.A2872359. A2933400.A2989549.A3307650		

MRDEF

(2004AB)

CUI	AUI	ATUI	SATUI	SAB	DEF	SUPPRESS	CVF
C0001403	A0019740	AT15061584		MSH	A disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin. It is due to tuberculosis- or autoimmune-induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. In the absence of replacement therapy, it is usually fatal.	N	

MRSAT (sample rows)

(2004AB)

	1	2	3	4	5	
	CUI	LUI	SUI	METAUI	STYPE	CODE
1	C0001403	L0001403	S0010792	A0019738	AUI	D000224
2	C0001403	L0001403	S0010794	A6326321	SCUI	C712
3	C0001403	L0001403	S0354372	A2922421	SAUI	363732003
4	C0001403			R15742591	SRUI	
5	C0001403				CUI	

	7	8	9	10	11	14	15
	ATUI	SATUI	ATN	SAB	ATV	SUPPRESS	CVF
1	AT15321482		DID	MSH	D000224	N	
2	AT33411754		MESH UI	NDFRT	D000224	N	
3	AT24166602		DESCRIPTION STATUS	SNOMEDCT	0	N	
4	AT27438950		REFINABILITY	SNOMEDCT	0	N	
5	AT02925340		ST	MTH	R	N	

MRSTY

(2004AB)

CUI	TUI	STN	STY	ATUI	CVF
C0001403	T047	B2.2.1.2.1	Disease or Syndrome	AT17683850	

MetamorphoSys

What is MetamorphoSys?

- Tool distributed with the UMLS
- Multi-platform Java software
- The UMLS installation and customization wizard
 - Installs Knowledge Sources to local storage
 - Subsets and customizes a local Metathesaurus
 - Unzips native Metathesaurus compressed files
- Simple to use
- Screens and tabs lead you through process

Customize the Metathesaurus

- Use MetamorphoSys
 - To remove unhelpful or harmful terminology
 - To comply with terms of license agreement
 - To change default settings
 - alter the preferred name
 - alter suppressibility of specific source term types
- Customization requires a clear understanding of:
 - Characteristics of source vocabularies
 - License arrangements
 - User's functional requirements, purpose and perspective
- Technical expertise requires multidisciplinary team

Machine Requirements

- A fast CPU – 1 GHz or higher
- 1 GB RAM recommended (512 MB min.)
- 6x (or better) DVD drive
- 13 GB minimum free disk space

- Runs on Sun Solaris 8 & 9, Windows XP, NT, and 2000, Linux, and Mac
- 1-10 hours run time on platforms tested
-- on average 4 hours or less

MetamorphoSys requires Java 1.5.

Download from UMLSKS ...

- High speed Internet connection required
- Read the README file for the release
- 2007AA UMLS Files
 - mmsys.zip (zipped MetamorphoSys application)
 - 2007AA-1-meta.nlm (compressed Metathesaurus data)
 - 2007AA-2-meta.nlm (compressed Metathesaurus data)
 - 2007AA-otherks.nlm (compressed Semantic Network and SPECIALIST Lexicon)
 - 2007AA.CHK
 - 2007AA.MD5
 - Copyright_Notice.txt
 - README.txt

...or DVD?

- Order at: custserv@nlm.nih.gov
- **Include your license number**

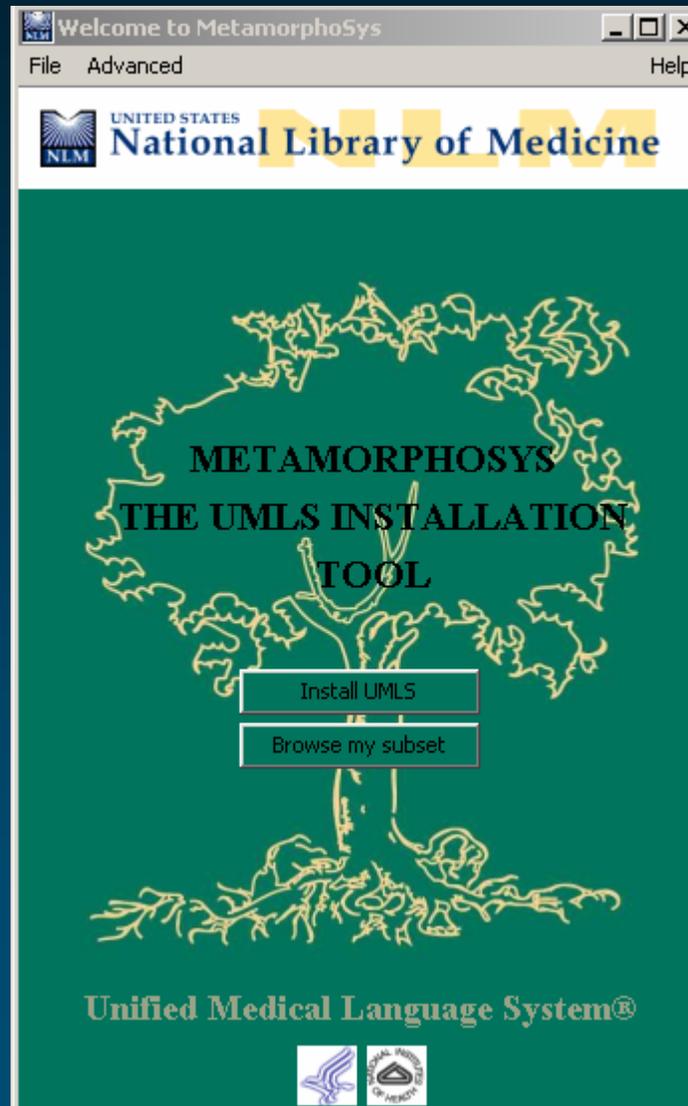
- Run MetamorphoSys from DVD
 - Windows
 - Autorun; or go to root directory and click on "windows_mmsys.bat"

 - Linux, Solaris, Macintosh
 - open a terminal window, change to the root directory and type appropriate command: `./linux_mmsys.sh`, `./solaris_mmsys.sh`, `./macintosh_mmsys.sh`

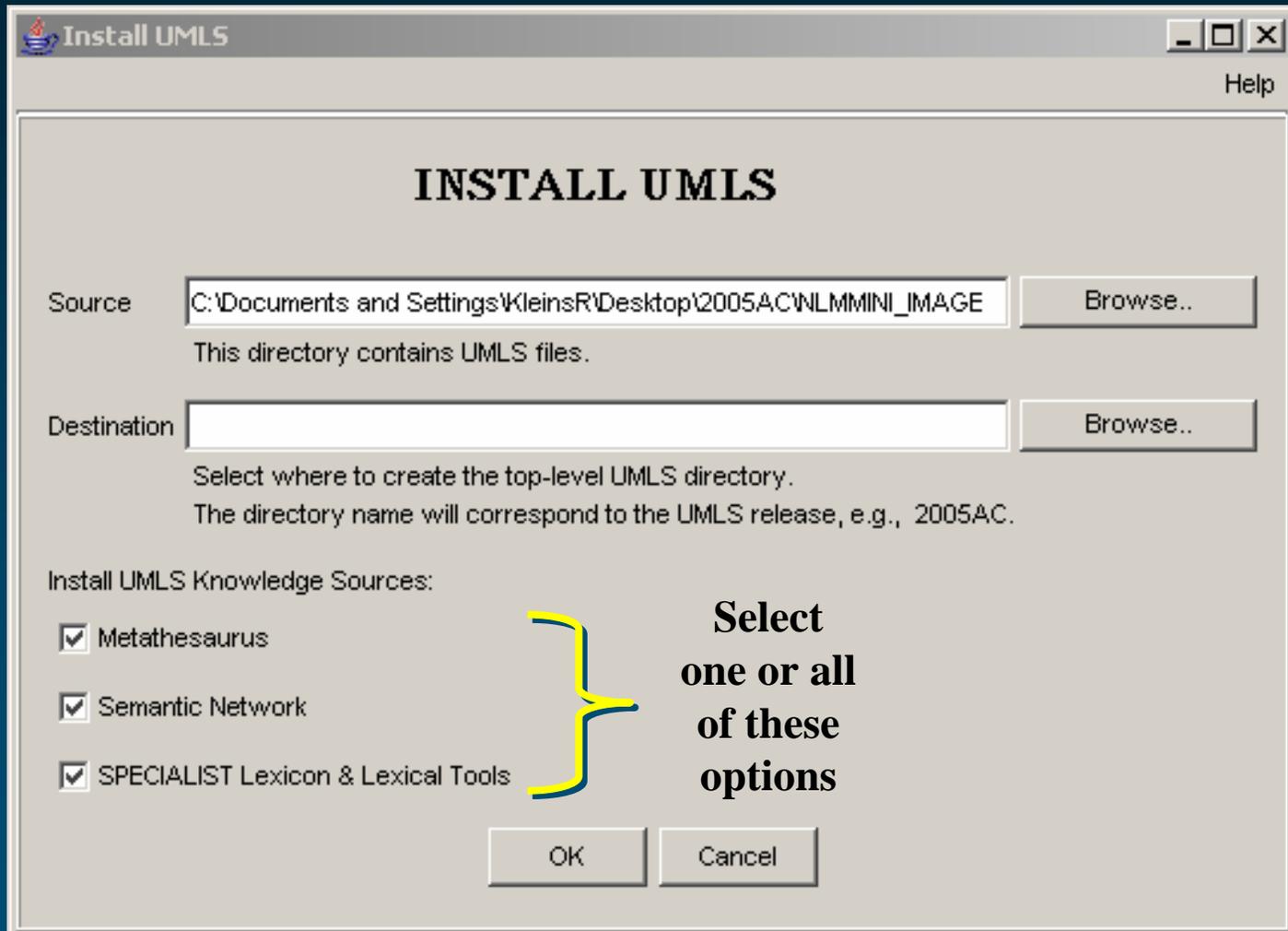
How MetamorphoSys Works

- Removes all information from all relational files from excluded vocabularies
 - atoms, strings, relationships, attributes, mappings
- Applies additional options selected by user
 - Changes to suppressibility or precedence
- Produces custom set of Metathesaurus relational files reflecting selected criteria
- Log file records subset details
- Output directory: set of Metathesaurus files

Welcome Screen



Install UMLS



Validation Summary

Validation Log

 **Validation Summary**

1862	files passed validation
0	files are missing
992	files have unexpected byte counts
0	files have unexpected content (the files may be corrupt or of the wrong version)

Validation started at: Wed Sep 12 16:24:37 EDT 2007
Validation ended at: Wed Sep 12 16:24:37 EDT 2007

The following file(s) have unexpected byte counts:

- Autorun.inf
- Copyright_Notice.txt
- MMSYS/config/att_types.dat
- MMSYS/config/mmsys.a.prop
- MMSYS/config/mmsys.b.prop
- MMSYS/config/mrpluscolsfiles.dat
- MMSYS/config/rel_types.dat
- MMSYS/config/snomed_rela_map.dat
- MMSYS/config/timestamp.dat

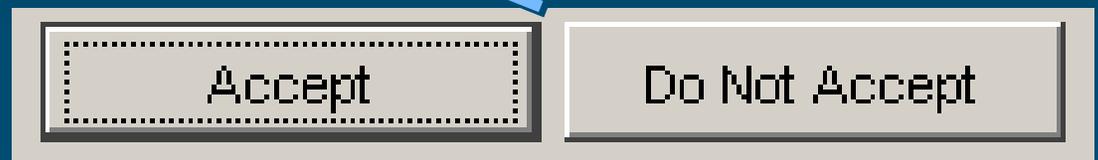
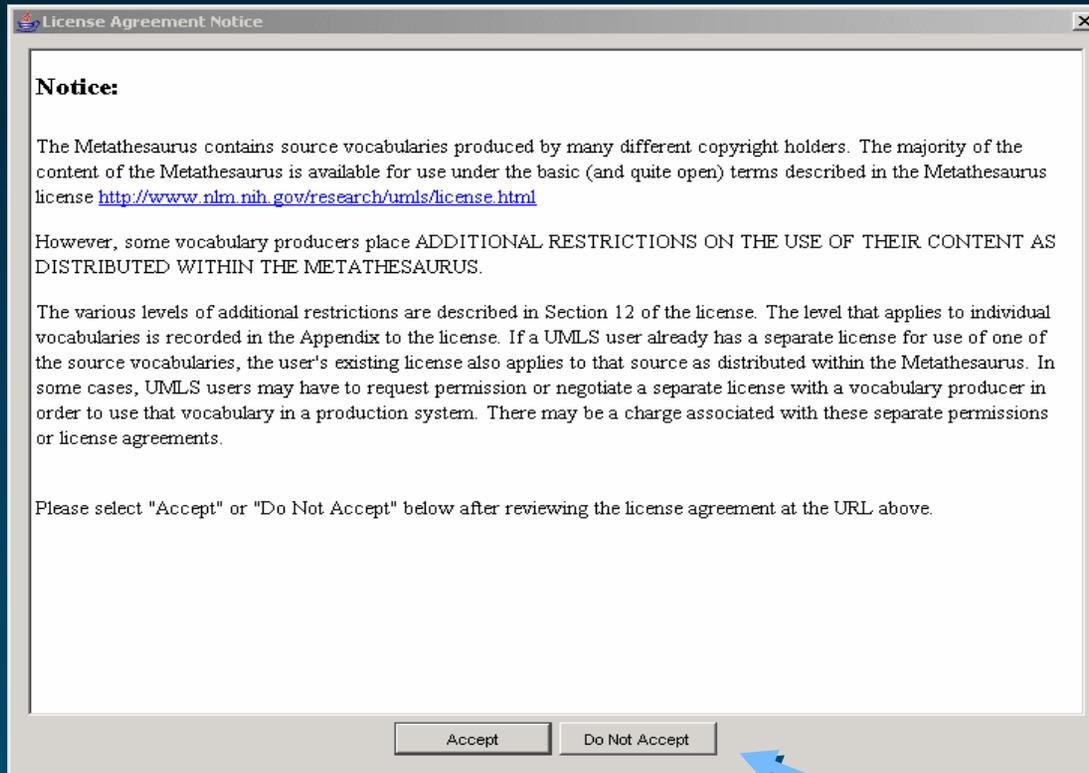
Continue Cancel

Configuration Files

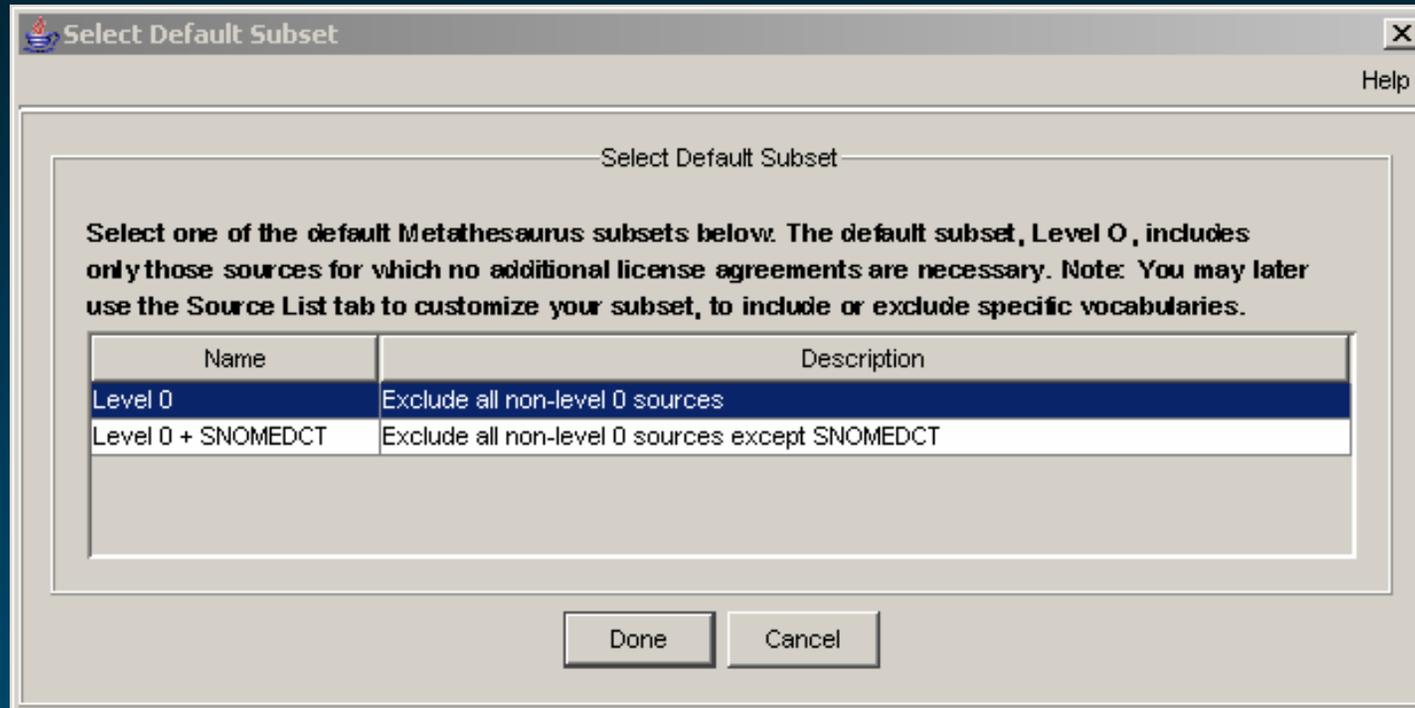


- New users must select "New Configuration"
- Returning users may select "Open Configuration" to open a previously saved MetamorphoSys Configuration
- This screen only appears if you install the Metathesaurus

UMLS License Notice



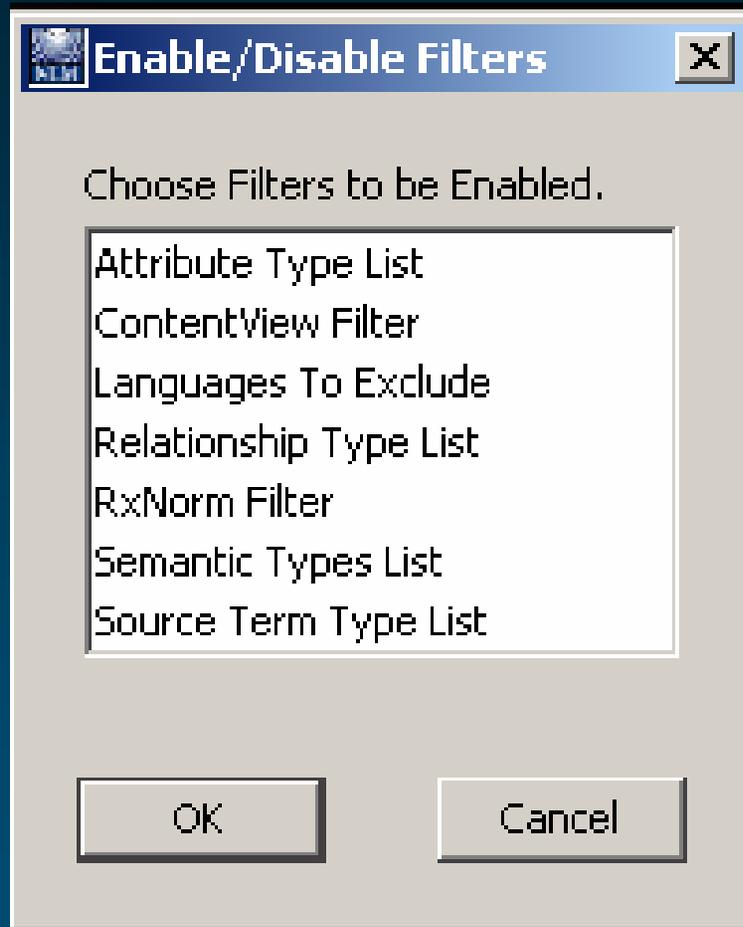
Select a default subset



Level 0 → no separate additional license agreements

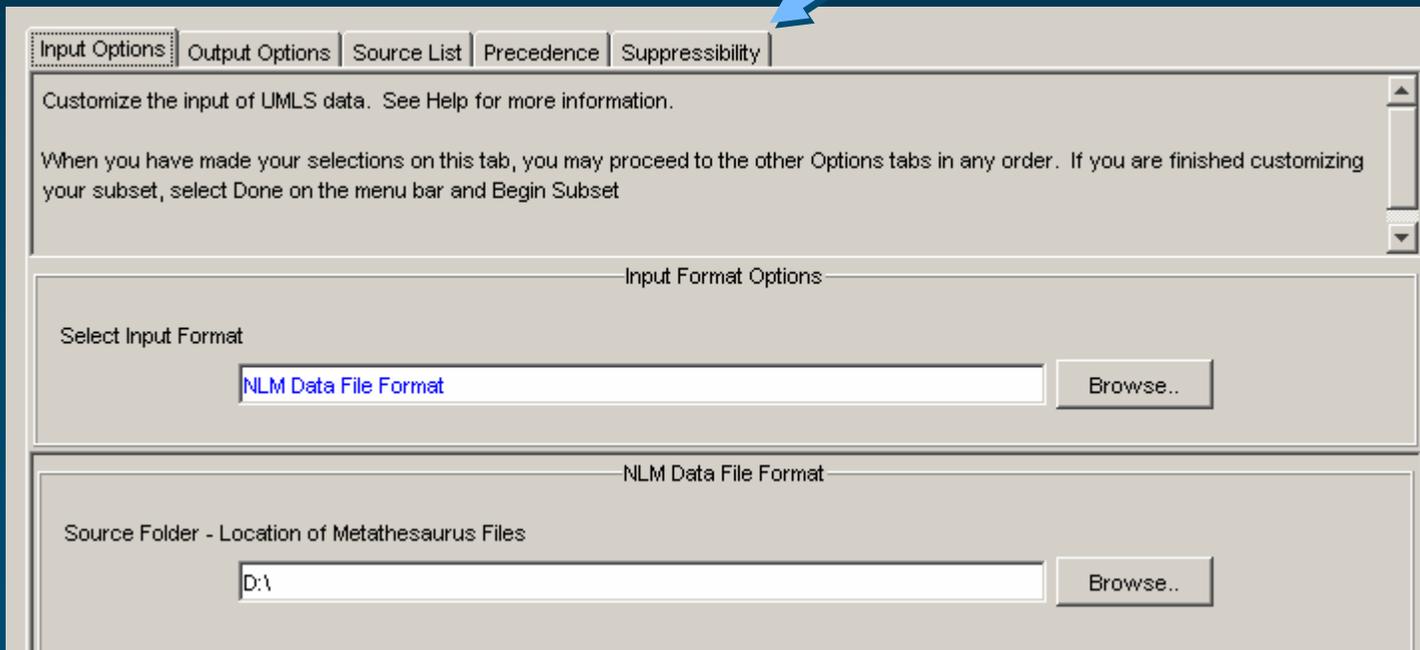
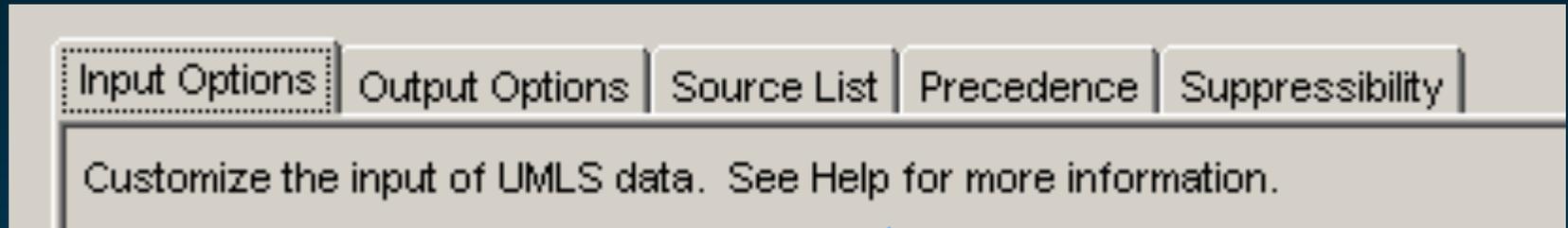
Level 0 + SNOMEDCT → Non-U.S. users must have separate license agreements

Filters



- Each filter enables an options tab
- Ctrl + Click to select more than one filter
- Several Filters available
 - Attributes by source
 - Languages
 - Relationships by source
 - Semantic Types
 - RxNorm
 - CVF (in development)

Input Options Tab



Output Options Tab

Input Options

Output Options

Source List

Precedence

Suppressibility

Select data output options for your local application. See Help for more information.

Calculate MD5s for output files - writes mmsys.md5 file.

Select Output Format

Subset Folder - Location of Subset Files

Remove records containing extended UTF-8 characters.

Truncate long fields to characters.

Output versioned source abbreviations rather than versionless source abbreviations.

Add UTF-8 BOM characters to output files.

Write Oracle load script.

Write Mysql load script.

Remove concepts containing only MTH atoms.

Source List Tab

Input Options | Output Options | **Source List** | Precedence | Suppressibility

Indicate below to INCLUDE or EXCLUDE selected sources.

SELECTED sources appear on a dark background, e.g., AIRRHEUM, 1993.

To undo selections and return to default source list, select "Reset Source List" from Reset menu at top.

Select sources to EXCLUDE from subset

Select sources to INCLUDE in subset

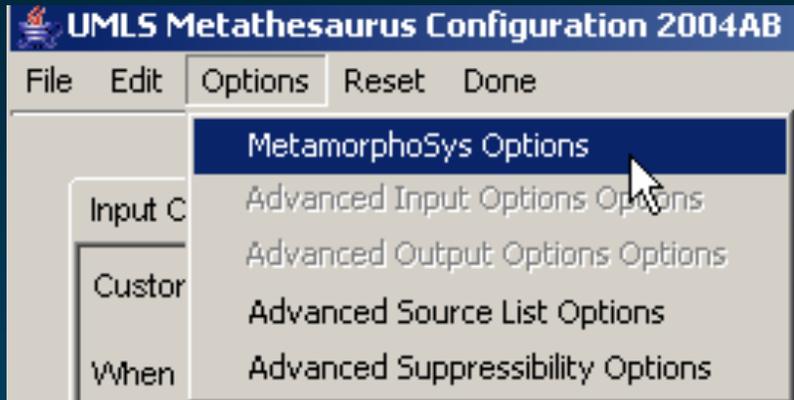
Exclude or Include

Sources to Exclude

Full Source Name	Source Abbreviation	Source Family	Language	Level
AIRRHEUM, 1993	AIR93	AIR	ENG	0
Alternative Billing Concepts, 2004	ALT2004	ALT	ENG	3
Alcohol and Other Drug Thesaurus, 2000	AOD2000	AOD	ENG	0
Beth Israel Vocabulary, 1.0	BI98	BI	ENG	2
Canonical Clinical Problem Statement System, 1999	CCPSS99	CCPSS	ENG	3
Clinical Classifications Software, 2003	CCS2003	CCS	ENG	0
Current Dental Terminology 2005 (CDT-5), 5	CDT5	CDT	ENG	3
COSTAR, 1989-1995	COSTAR_89-95	COSTAR	ENG	0
Medical Entities Dictionary, 2003	CPM2003	CPM	ENG	2
Physicians' Current Procedural Terminology, Spanish Translation, 2001	CPT01SP	CPT	SPA	3
Physicians' Current Procedural Terminology, 2005	CPT2005	CPT	ENG	3
CRISP Thesaurus, 2004	CSP2004	CSP	ENG	0
COSTART, 1995	CST95	CST	ENG	0
Diseases Database, 2000	DDB00	DDB	ENG	3
German translation of ICD10, 1995	DMDICD10_1995	ICD10	GER	1
German translation of UMDNS, 1996	DMDUMD_1996	UMD	GER	1
DSM-III-R, 1987	DSM3R_1987	DSM3R	ENG	3
DSM-IV, 1994	DSM4_1994	DSM4	ENG	3
DXplain, 1994	DXP94	DXP	ENG	0
Gene Ontology, 2004_12_20	GO2004_12_20	GO	ENG	0
HCPCS Version of Current Dental Terminology 2005 (CDT-5), 5	HCDT5	CDT	ENG	3

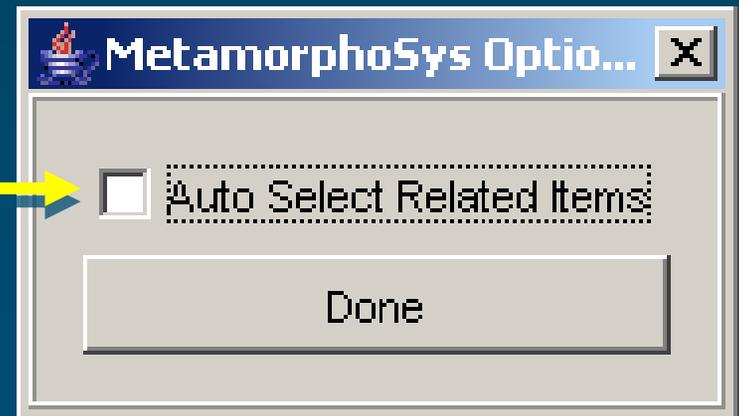
Highlighted rows are excluded from the subset.

MetamorphoSys Option Tab



Source list behavior can be changed using the MetamorphoSys Option Tab

If you wish to Auto Select Related Items check this box



Precedence Tab



- Ranks names by types of terms within sources
- Highest ranking name determines the Preferred Name

Input Options | Output Options | Source List | **Precedence** | Suppressibility

Change the ranking of sources and their associated term types to create concept names that are more useful in your local application. Concept names are determined by the term with highest ranking source/term type.

To move rows, either cut and paste rows, or drag and drop.

When you have made your selections on this tab, you may proceed to the other Options tabs in any order. If you are finished customizing

Precedence

Full Source Name	Source Abbreviation	Term Type
UMLS Metathesaurus	MTH	PN
Medical Subject Headings, MSH2004_2003_12_12	MSH2004_2003_12_12	MH
Medical Subject Headings, MSH2004_2003_12_12	MSH2004_2003_12_12	TQ
Medical Subject Headings, MSH2004_2003_12_12	MSH2004_2003_12_12	EP
Medical Subject Headings, MSH2004_2003_12_12	MSH2004_2003_12_12	EN
Medical Subject Headings, MSH2004_2003_12_12	MSH2004_2003_12_12	XQ
Medical Subject Headings, MSH2004_2003_12_12	MSH2004_2003_12_12	NM
RXNORM Project, META2004AB	RXNORM_04AB	SCD

Cut and paste rows to alter the preferred name

Suppressibility Tab



Input Options | Output Options | Source List | Precedence | **Suppressibility**

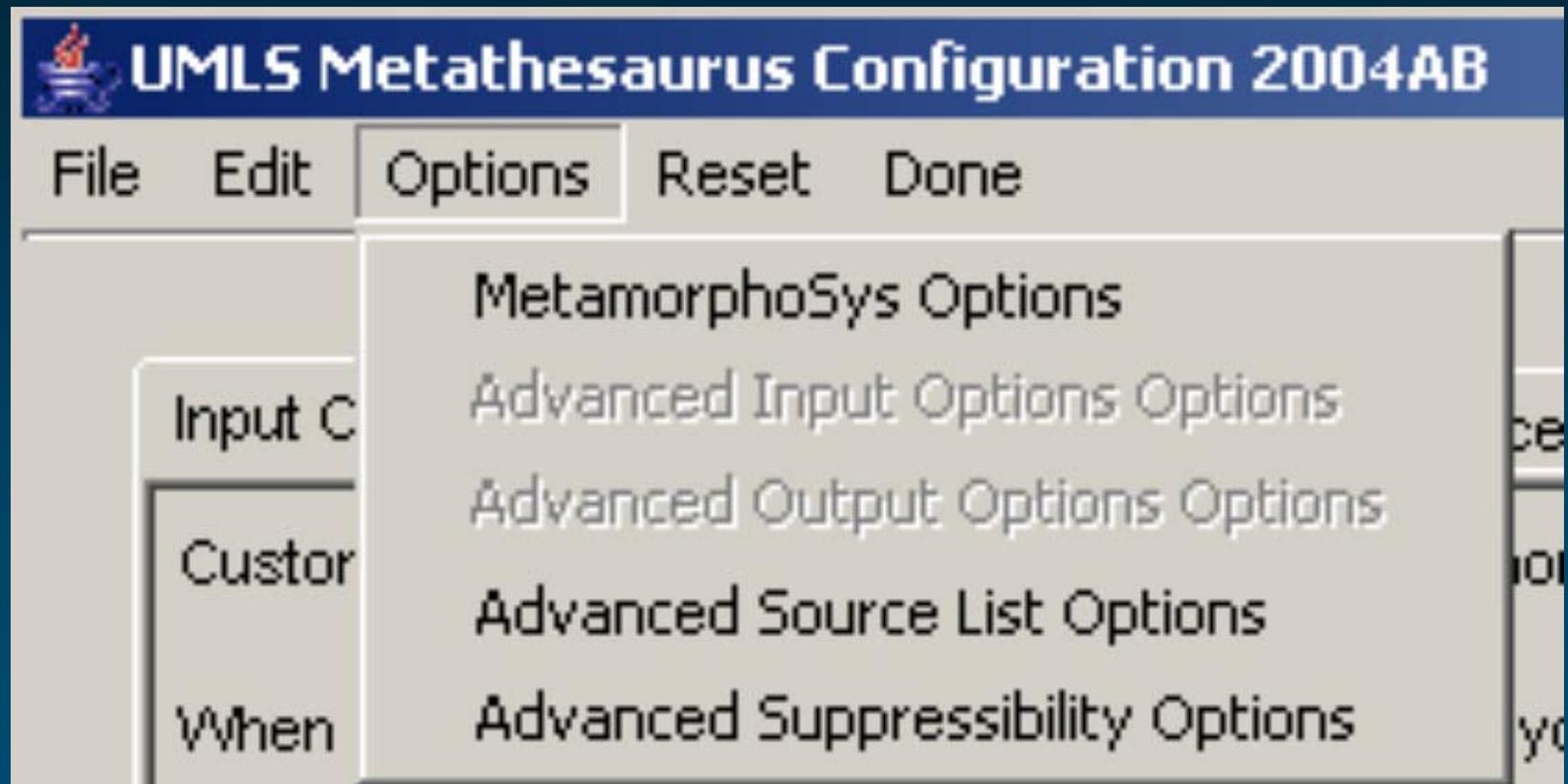
See Help for more information.

When you have made your selections on this tab, you may proceed to the other Options tabs in any order. If you are finished customizing your subset, select Done on the menu bar and Begin Subset.

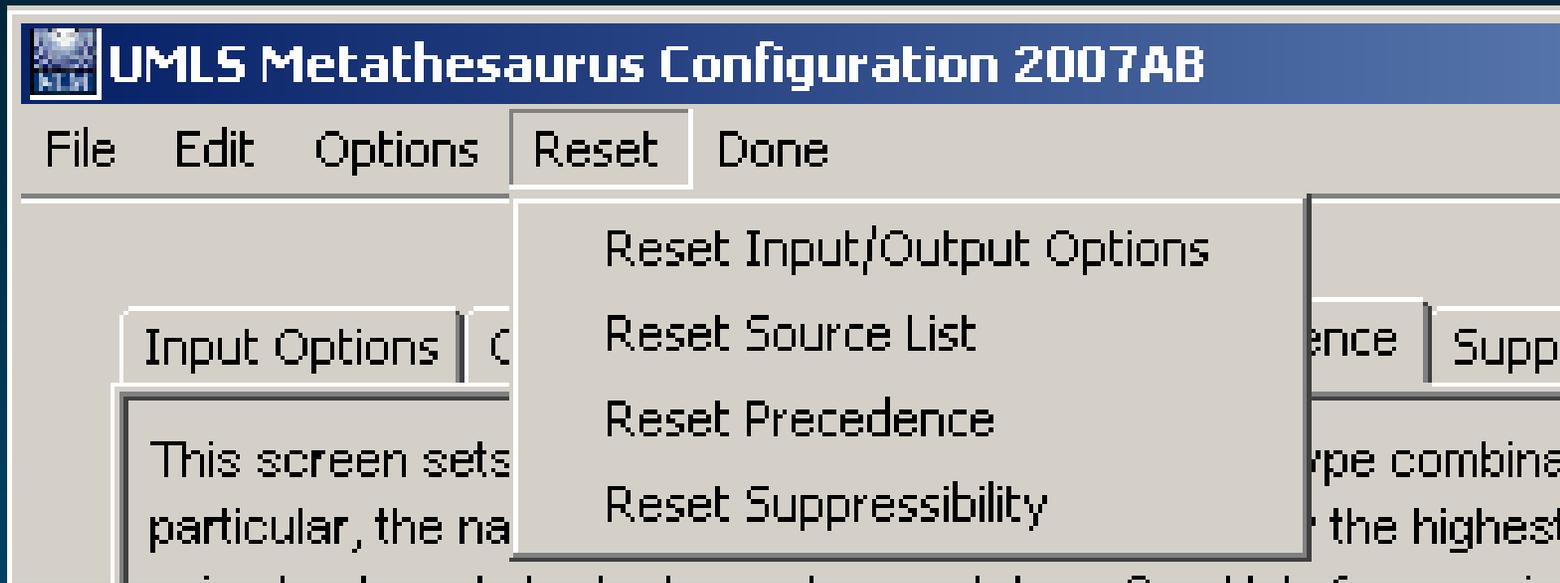
International Classification of Primary Care, 1993	ICPC93	CP
International Classification of Primary Care, 1993	ICPC93	CS
International Classification of Primary Care, 1993	ICPC93	CX
International Classification of Primary Care, 1993	ICPC93	HT
International Classification of Primary Care, 1993	ICPC93	PC
International Classification of Primary Care, 1993	ICPC93	PS
International Classification of Primary Care, 1993	ICPC93	PT
International Classification of Primary Care, 1993	ICPC93	PX
ICPC, Basque Translation, 1993	ICPCBAQ_1993	CP

Highlighted source term types will be marked suppressible

Options menu



Reset menu

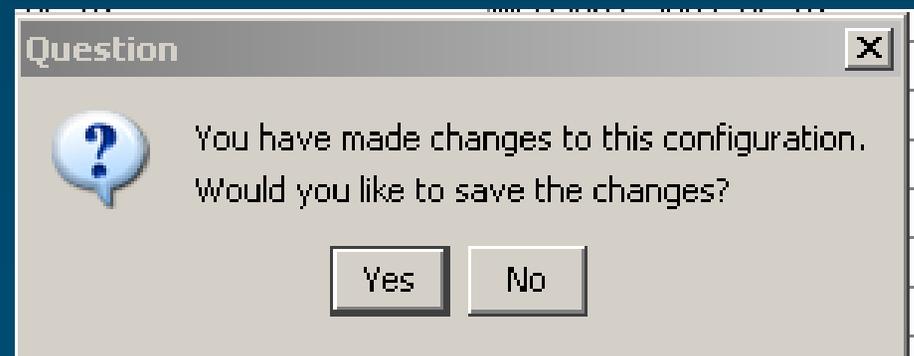


- Returns all filters to default selections
- Default selections in "mmsys.prop.default file" in config folder
- mmsys.prop.default contains properties in last run

Done – Begin Subset



- Complete configuration options
- Done menu
- Begin Subset



MetamorphoSys log

mmsys.log - Notepad

File Edit Format View Help

```
MetamorphoSys Version:.....5.21
MetamorphoSys Build Date:.....2004_08_30_14_47_11
UMLS Build Date:.....2004_07_12_09_57_26
Release Version:.....2004AB
Release Date:.....20040720
Release Description:.....July 2004 release
Metathesaurus source paths:.....C:\UMLS\DVDIMAGE
Subsetted Metathesaurus folder:.....C:\UMLS\DVDIMAGE\2004AB\META
Start at:.....Wed Sep 01 13:04:18 EDT 2004
Initialize CUI List completed:.....Wed Sep 01 13:06:20 EDT 2004
Subset Metathesaurus completed:.....Wed Sep 01 14:13:02 EDT 2004
Subset Index Files completed:.....Wed Sep 01 14:21:11 EDT 2004
Subset Release Metadata completed:.....Wed Sep 01 14:21:17 EDT 2004
Finished at:.....Wed Sep 01 14:22:48 EDT 2004
Concepts in source:.....1078246
Concepts in subset:.....1078246
Time elapsed:.....01:18:29
```

MetamorphoSys log

mmsys.log - Notepad

File Edit Format View Help

Metathesaurus Output: Rich Release Format

Long fields were not truncated.

Source Abbreviations were written out with a versionless (root) representation.

Fields containing UTF-8 characters were not removed.

Excluded Sources

<none>

Kept Sources

AI/RHEUM, 1993	AIR93
Alternative Billing Concepts	ALT2003
Alcohol and Other Drug Thesaurus, 2000	AOD2000
Beth Israel Vocabulary, 1.0	BI98
Canonical Clinical Problem Statement System, 1999	CCPSS99
Clinical Classifications software, 2003	CCS2003
Current Dental Terminology (CDT), 4	CDT4
COSTAR, 1989-1995	COSTAR_89-95
Medical Entities Dictionary, 2003	CPM2003
Physicians' Current Procedural Terminology, spanish translation,...	CPT01SP
Physicians' Current Procedural Terminology, 2004	CPT2004
CRISP Thesaurus, 2004	CSP2004
COSTART, 1995	CST95
Diseases Database, 2000	DDB00
German translation of ICD10, 1995	DMDICD10_1995
German translation of UMDNS, 1996	DMDUMD_1996

Output directory contents

Name	Size	Type ▲
 CHANGE		File Folder
 indexes		File Folder
 release.dat	1 KB	DAT File
 config.prop	8 KB	PROP File
 AMBIGLUI.RRF	1,225 KB	RRF File
 AMBIGSUI.RRF	955 KB	RRF File
 MRCOC.RRF	809,207 KB	RRF File
 MRCOLS.RRF	21 KB	RRF File
 MRCONSO.RRF	596,528 KB	RRF File
 MRCUI.RRF	9,221 KB	RRF File
 MRCXT.RRF	9,391,778 KB	RRF File
 MRDEF.RRF	17,172 KB	RRF File
 MRDOC.RRF	88 KB	RRF File
 MRFILES.RRF	4 KB	RRF File
 MRHIER.RRF	899,786 KB	RRF File
 MRHIST.RRF	70,843 KB	RRF File
 MRMAP.RRF	9,362 KB	RRF File

RRF Subset Browser

- Must have an RRF Subset
- View tree, report, or raw data for each term
- Search for CUI or more information
- Quickly find terms within a subset

http://www.nlm.nih.gov/research/umls/rrf_help.html

RRF Subset Browser

- View customized subsets
- Expandable Concept Report View
- Raw View: relational table data
- Hyperlinked concepts

RRF Browser word search & report view

The screenshot shows the 'Rich Release Format Browser 2005AB C0001403' application window. The interface includes a menu bar (File, Edit, Options, Help), a toolbar with navigation icons, and a 'Subset Directory' field showing the path 'C:\Documents and Settings\Jan Willis\My Documents\Old N...'. The main area is divided into two panes: 'Word Search' and 'Report View'.

Word Search Panel:

- Buttons: CUI Search, Tree Browser, Word Search
- Text: Enter search terms: (English)
- Input field: addison's disease
- Button: Search..
- Text: Select a concept. (1 to 12 of 15 results)
- Navigation: Left and right arrow buttons
- Results list:
 - C0001403 Primary adrenocortical insufficiency
 - C0271737 Addison's disease due to autoimmunity
 - C0546992 Tuberculous Addison's disease
 - C0342477 Addison's disease with adrenoleucodystrophy
 - C0270989 Myopathy in Addison's disease
 - C0268785 Salt-losing nephropathy
 - C0271739 Familial adrenocortical hypoplasia
 - C0002892 Pernicious anemia
 - C0036420 Unspecified circumscribed scleroderma
 - C0271740 Congenital primary adrenocortical hypofunction
 - C0085859 Polyglandular autoimmune syndrome, type 1
 - C0085860 Polyglandular autoimmune syndrome, type 2

Report View Panel:

- Buttons: Raw View, Report View
- Content:
 - Concept: [CUI: C0001403]** Primary adrenocortical insufficiency
 - Semantic Type**
 - Disease or Syndrome
 - Atoms (5):** (Sorted by Source, String)
 - Contexts (40)**
 - SNOMEDCT/PT/363732003 10 Addison's disease
 - ANC**
 - [SNOMED CT Concept](#)
 - [Clinical finding](#)
 - [Finding by site](#)
 - [Finding of body region](#)
 - [Finding of trunk structure](#)
 - [General finding of abdomen](#)
 - [Disorder of abdomen](#)
 - [Disorder of adrenal gland](#)
 - [Disorder of adrenal cortex](#)
 - [Adrenal cortical hypofunction](#)
 - SIB**
 - CHD**
 - [Addison's disease due to autoimmunity](#)
 - [Addison's disease with adrenoleucodystrophy](#)
 - [Polyglandular autoimmune syndrome, type 1](#)
 - [Tuberculous Addison's disease](#)
 - SNOMEDCT/PT/363732003 11 Addison's disease

RRF Browser tree browser & report view

The screenshot displays the 'Rich Release Format Browser 2005AB C0001403' application window. The interface includes a menu bar (File, Edit, Options, Help), a toolbar with navigation icons, and a 'Subset Directory' field showing the path 'C:\Documents and Settings\Jan Willis\My Documents\Old No...'. The main area is divided into two panes: 'Tree Browser' and 'Report View'.

Tree Browser: Shows a 'Context Tree Tops' list with expandable items:

- [-] SNOMED CT Concept
 - [+] Attribute
 - [+] Body structure
 - [+] Clinical finding
 - [+] Context-dependent categories
 - [+] Environments and geographical locations
 - [+] Events
 - [+] Observable entity
 - [+] Organism
 - [+] Pharmaceutical / biologic product
 - [+] Physical force
 - [+] Physical object
 - [+] Procedure
 - [+] Qualifier value
 - [+] Social context
 - [+] Special concept
 - [+] Specimen
 - [+] Staging and scales
 - [+] Substance

Report View: Displays details for the selected concept:

- [+] **Concept: [CUI: C0001403]** Primary adrenocortical insufficiency
- [-] **Semantic Type** Disease or Syndrome
- [+] **Atoms (5):** (Sorted by Source, String)
- [-] **Contexts (40)**
 - [-] SNOMEDCT/PT/363732003 10 Addison's disease
 - [-] **ANC**
 - [SNOMED CT Concept](#)
 - [Clinical finding](#)
 - [Finding by site](#)
 - [Finding of body region](#)
 - [Finding of trunk structure](#)
 - [General finding of abdomen](#)
 - [Disorder of abdomen](#)
 - [Disorder of adrenal gland](#)
 - [Disorder of adrenal cortex](#)
 - [Adrenal cortical hypofunction](#)
 - [+] **SIB**
 - [-] **CHD**
 - [Addison's disease due to autoimmunity](#)
 - [Addison's disease with adrenoleucodystrophy](#)
 - [Polyglandular autoimmune syndrome, type 1](#)
 - [Tuberculous Addison's disease](#)

RRF Browser -- search by code

The screenshot shows the 'Rich Release Format Browser' application window. The title bar reads 'Rich Release Format Browser 2007AB-AC-20070904 C0001002'. The menu bar includes 'File', 'Edit', 'Options', and 'Help'. The address bar shows the file path: 'C:\Documents and Settings\kleinsr\Desktop\test rxnorm\2007AB-AC-20070904\M...'. The interface has several tabs: 'CUI Search', 'Code Search', 'Tree Browser', and 'Word Search'. The 'Code Search' tab is active, showing a search input field with 'D000096' and a 'Get Concepts..' button. Below this, it says 'Select a concept. (1 result)' with left and right arrow buttons. A list box shows 'C0001002 Acetone' selected. To the right, there are 'Raw View' and 'Report View' tabs. The 'Raw View' tab is active, displaying a structured text output for the selected concept. The output includes sections for 'Concept', 'Semantic Types', 'Definitions', and 'Atoms'.

Rich Release Format Browser 2007AB-AC-20070904 C0001002

File Edit Options Help

Monosp... ..

C:\Documents and Settings\kleinsr\Desktop\test rxnorm\2007AB-AC-20070904\M...

CUI Search Code Search Tree Browser Word Search

Enter a CODE:

D000096

Get Concepts..

Select a concept. (1 result)

C0001002 Acetone

Raw View Report View

- ⊕ **Concept:** [CUI C0001002] Acetone
- ⊖ **Semantic Types**
 - Organic Chemical
 - Pharmacologic Substance
- ⊖ **Definitions**

MSH/MH|A colorless liquid used as a solvent and an antiseptic. It is one of the ketone bodies produced during ketoacidosis.

MSHSWE/MH|En brandfarlig, färglös, flyktig vätska med behaglig eterisk doft. Den är ett allmänt använt lösningsmedel och används som sådant inom farmacin; vid koncentrationer över 80% används den som antiseptis kt medel. Aceton är ett av de ketonämnen som bildas vid ketoacidosis, som i diabetes eller vid svält.
- ⊖ **Atoms (16):** (Sorted by Source, String)
 - 2-Propanone [A0527815/MSH/NL/D000096]
 - ⊕ Acetone [A0018351/MSH/MH/D000096]
 - ACETON [A3910037/MSHCZE/MH/D000096]
 - Asetoni [A9072482/MSHFIN/MH/D000096]
 - Acétone [A7439779/MSHFRE/MH/D000096]
 - Aceton [A7481179/MSHGER/MH/D000096]
 - Azeton [A7483923/MSHGER/EP/D000096]

UMLS
Knowledge Source Server
(UMLSKS)

UMLS Knowledge Source Server

- Licensed users access online:
<http://umlsks.nlm.nih.gov>
- Web search interface for the three Knowledge Sources
- Application Programming Interface (API)

UMLSKS Welcome page

UMLS Knowledge Source Server (UMLSKS)
National Library of Medicine

About the UMLSKS

- Learn
- Overview
- Frequently Asked Questions
- UMLS Metathesaurus®
- Semantic Network
- SPECIALIST Lexicon

Documentation

- User's Guide
- Developer's Guide
- UMLS Documentation Set

UMLSKS Registered Users

Login ID:
Password:
Login
[Forgotten your password?](#)

New UMLSKS Users

The UMLSKS is restricted to registered users. If you are not a registered user and would like to use the resources of the UMLS, please read the [UMLS license agreement](#), obtain a UMLS license number, and fill in the form below to request a UMLSKS membership.

Login ID:
Password:
Confirm Password:
First Name:
Last Name:
Request Account

What's New

- UMLSKS Version 4.2 released on 07/12/04. Look [here](#) for a list of new features and bug fixes.
- 2004 AA Metathesaurus – including SNOMED CT® – now available to download for those that have signed the new license agreement!
- Existing users may click on the [Forgotten your password link](#) (above), and enter the user license

Document: Done (0.845 secs)

- Returning users log in

- New users create account

UMLS Knowledge Source Server Home Page

- Tabs across top access basic searching of 3 Knowledge Sources
- Advanced searching options on right-hand side

Netscape: UMLS Knowledge Source Server (UMLSKS)

File Edit View Go Communicator Help

Back Forward Reload Home Search Netscape Print Security Shop Stop

Bookmarks Location: <http://umlsks.nlm.nih.gov/kss/servlet/Turbine/act/> What's Related

Members WebMail Connections BizJournal SmartUpdate Mktplace

UMLS Knowledge Source Server (UMLSKS)

UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

Metathesaurus **Semantic Network** **SPECIALIST Lexicon**

Logout

About the UMLSKS

- ▶ [Home](#)
- ▶ [Overview](#)
- ▶ [Frequently Asked Questions](#)
- ▶ [Edit Views/Profile](#)

Downloads

- ▶ [UMLS Knowledge Sources](#)
- ▶ [Developer's API](#)

Documentation

- ▶ [User's Guide](#)
- ▶ [Developer's Guide](#)
- ▶ [Developer's API Javadocs](#)
- ▶ [UMLS Documentation Set](#)

Resources

- ▶ [NLP & Lexical Resources](#)
- ▶ [Semantic Network Resources](#)
- ▶ [Metathesaurus Resources](#)

Quick Search

Select UMLS Release:

Enter search value:

[Metathesaurus Search](#) [Semantic Network Search](#) [SPECIALIST Lexicon Search](#)

[Search Tips...](#) [Search Tips...](#) [Search Tips...](#)

Advanced Searches

[Metathesaurus Advanced Search](#)

Facilitates advanced searching of the UMLS Metathesaurus, including restricting vocabularies, performing batch searches, performing XML queries, and using a command-line type interface.

[Semantic Network Browser](#)

Allows browsing of the hierarchies for the Semantic Network.

What's New

- ▶ 2004 AB Metathesaurus now available to download and searching for those that have signed the new license agreement!
- ▶ UMLSKS Version 4.3 released on August 30, 2004 for 2004 AB download access and searching.

Metathesaurus Basic Search

Addison's disease

The screenshot displays the UMLS Knowledge Source Server (UMLSKS) website. The header includes the UMLSKS logo, version information (UMLSKS Version 4.2, UMLS Releases: 2002, 2002AB, 2002AC, 2002AD, 2003AA, 2003AB, 2003AC, 2004AA), and navigation links for Metathesaurus, Semantic Network, and SPECIALIST Lexicon. A 'Logout' link is also present.

The main content area is divided into several sections:

- Quick Search:** A dropdown menu for 'Select UMLS Release:' is set to '2004AA'. Below it is a text input field for 'Enter search value:'.
- Advanced Searches:** A link for 'Metathesaurus Advanced Search' is visible, with a description: 'Facilitates advanced searching of the UMLS Metathesaurus including restricting...'
- Search Options:** Three red buttons are displayed: 'Metathesaurus Concept Search', 'Semantic Network Search', and 'SPECIALIST Lexicon Search'. Each button has a corresponding 'Search Tips...' link below it.

A search box is highlighted with a black border, showing the text 'Addison's disease' entered in the 'Enter search value:' field. The 'Select UMLS Release:' dropdown is also highlighted, showing '2004AB' selected.

- UMLS Release
- Search Term
- UMLS Knowledge Source

UMLS Knowledge Source Server (UMLSKS) Basic Concept Report

The screenshot displays the UMLS Knowledge Source Server (UMLSKS) interface. At the top, there is a logo of a tree and the text "UMLS Knowledge Source Server (UMLSKS)". Below this, it says "UMLSKS Version 4.2.2" and "UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB". The main navigation bar includes "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". There are links for "Home", "Advanced Search", and "Logout".

The search results section shows a search for "addison's disease" in UMLS Release 2004AB. There are two buttons: "Display" and "Display All".

Concept: Addison's disease
CUI: C0001403
Semantic Type: Disease or Syndrome

Definition:
A disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin. It is due to tuberculosis- or autoimmune-induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. In the absence of replacement therapy, it is usually fatal. (MeSH)

disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin; due to tuberculosis or autoimmune induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. (CRISP Thesaurus)

Synonyms:
Addison's disease
Addison's disease (disorder)
ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)
ADRENOCORTICAL INSUFFICIENCY, PRIMARY
Asthenia pigmentosa
Bronzed disease

On the left side, there are sections for "Concept", "Context", and "Relations", each with several checkboxes. Under "Concept", "Definition" and "Synonyms" are checked. Under "Context", "Ancestors", "Parents", "Siblings", and "Children" are unchecked. Under "Relations", "Narrower", "Broader", "Similar", and "Other" are unchecked, while "Related and possibly synonyms" is checked.

● Concept Name/
CUI

● Semantic
Type(s)

● Definition(s)

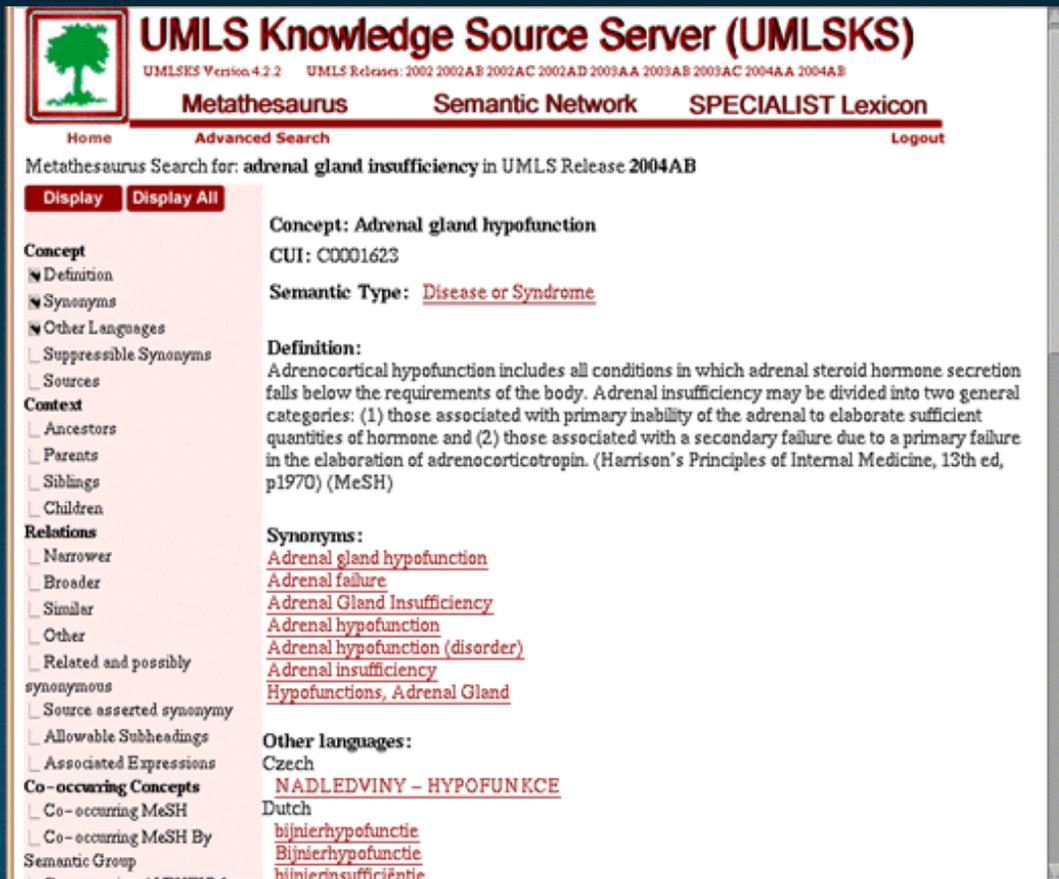
● Synonyms

Display All

The screenshot shows the UMLS Knowledge Source Server (UMLSKS) interface. At the top, there is a logo of a tree and the text "UMLS Knowledge Source Server (UMLSKS)" with version and release information. Below this are navigation links for "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". A search bar contains the text "Metathesaurus Search for: addison's disease in UMLS Release 2004AB". Two red buttons, "Display" and "Display All", are visible. A black box highlights the "Display All" button in the search results area. A blue arrow points from the "Display All" button in the search results to the "Display All" button in the search bar. Another blue arrow points from the "Display All" button in the search bar to the "Display All" button in the search results. The search results for "Addison's disease" (CUI: C0001403) are shown, including a definition, synonyms, and a list of related terms such as "ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)", "ADRENOCORTICAL INSUFFICIENCY, PRIMARY FAILURE", "Asthenia pigmentosa", and "Bronzed disease".

- “Display” shows results for selected options
- “Display All” shows results for all available options

Concept Report Display All *Adrenal Gland Insufficiency*



UMLS Knowledge Source Server (UMLSKS)
UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

Metathesaurus Semantic Network SPECIALIST Lexicon

Home Advanced Search Logout

Metathesaurus Search for: **adrenal gland insufficiency** in UMLS Release 2004AB

Display **Display All**

Concept
 Definition
 Synonyms
 Other Languages
 Suppressible Synonyms
 Sources

Context
 Ancestors
 Parents
 Siblings
 Children

Relations
 Narrower
 Broader
 Similar
 Other
 Related and possibly
synonyms
 Source asserted synonymy
 Allowable Subheadings
 Associated Expressions

Co-occurring Concepts
 Co-occurring MeSH
 Co-occurring MeSH By
Semantic Group

Concept: Adrenal gland hypofunction
CUI: C0001623

Semantic Type: [Disease or Syndrome](#)

Definition:
Adrenocortical hypofunction includes all conditions in which adrenal steroid hormone secretion falls below the requirements of the body. Adrenal insufficiency may be divided into two general categories: (1) those associated with primary inability of the adrenal to elaborate sufficient quantities of hormone and (2) those associated with a secondary failure due to a primary failure in the elaboration of adrenocorticotropin. (Harrison's Principles of Internal Medicine, 13th ed, p1970) (MeSH)

Synonyms:
[Adrenal gland hypofunction](#)
[Adrenal failure](#)
[Adrenal Gland Insufficiency](#)
[Adrenal hypofunction](#)
[Adrenal hypofunction \(disorder\)](#)
[Adrenal insufficiency](#)
[Hypofunctions, Adrenal Gland](#)

Other languages:
Czech
[NADLEDVINY – HYPOFUNKCE](#)
Dutch
[bijnierhypofunctie](#)
[Bijnierhypofunctie](#)
[hijnierinsufficiëntie](#)

- Concept Name/CUI
- Semantic Type(s)
- Definition(s)
- Synonyms, including foreign languages
- Relations (broader, narrower, etc.)
- Co-occurrence data

Concept Report Display All (continued)

Synonyms

Sources

The screenshot displays a list of synonyms for a medical concept, organized by language. The languages listed are Japanese, Portuguese, Russian, Spanish, and Swedish. The synonyms are underlined and include terms like '副腎機能低下', 'HIPOFUNCAO SUPRA-RENAL', 'НАДПОЧЕЧНИКОВ ГИПОФУНКЦИЯ', and 'hipofuncion adrenal'. Below the synonyms, there is a section for 'Suppressible Synonyms' and a list of 'Sources' including Beth Israel Problem List, Clinical Problem Statements, COSTAR, CRISP Thesaurus, and COSTART.

Japanese
副腎機能低下
副腎機能低下症

Portuguese
Hipofunção das Glândulas Supra-Renais
HIPOFUNCAO SUPRA-RENAL
Hipofunção supra-renal
INSUFICIENCIA SUPRA-RENAL
Insuficiência supra-renal

Russian
НАДПОЧЕЧНИКОВ ГИПОФУНКЦИЯ

Spanish
hipofuncion adrenal
hipofunción adrenal
Hipofunción de las Glándulas Suprarrenales
hipofuncion suprarrenal
Hipofunción suprarrenal
hipofunción suprarrenal
Hipofunción suprarrenal
hipofuncion suprarrenal
hipofunción suprarrenal
Insuficiencia adrenal
Insuficiencia supra-renal
INSUFICIENCIA SUPRARRENAL
Insuficiencia suprarrenal
SUPRARRENAL HIPOFUNCION

Swedish
Binjurehypofunktion

Suppressible Synonyms:
adrenal insufficiency <2>

Sources:
Beth Israel Problem List
Clinical Problem Statements
COSTAR
CRISP Thesaurus
COSTART

Concept Report Display All (continued)

Hierarchies

The screenshot displays a concept report with the following sections:

- Ancestors:**
 - MeSH**
 - [MeSH Descriptors \[\]](#)
 - [Index Medicus Descriptor \[\]](#)
 - [Diseases \(MeSH Category\) \[C\]](#)
 - [Endocrine Diseases \[C19\]](#)
 - [Adrenal Gland Diseases \[C19.053\]](#)
 - [Adrenal Gland Hypofunction \[C19.053.264\]](#)
 - MedDRA**
 - [Endocrine dis](#)
 - [Adrenal glan](#)
 - [Adrenal co](#)
 - [Adrenal in](#)
 - MedDRA**
 - [Metabolism a](#)
 - [Metabolism](#)
 - [Metabolic c](#)
 - [Adrenal](#)
 - NCI Thesaur**
 - [Diseases, Disorders and Findings \[\]](#)
 - [Diseases and Disorders \[\]](#)
 - [Disorder by Site \[\]](#)
 - [Endocrine Disorder \[\]](#)
 - [Adrenal Gland Disorder \[\]](#)
 - [Non-Neoplastic Adrenal Gland Disorder \[\]](#)
 - [Adrenal Gland Insufficiency \[\]](#)
 - NCI Thesaurus**
 - [Diseases, Disorders and Findings \[\]](#)
 - [Diseases and Disorders \[\]](#)
 - [Disorder by Site \[\]](#)

A callout box highlights the MeSH hierarchy, showing the same list of terms as above.

Concept Report Display All (continued)

THYROIDITIS [0420]
THYROXINE DECREASED [1349]
TSH DECREASED [1690]
TSH INCREASED [1831]
VIRILISM [0421]

Narrower Concepts:

Addison's disease (National Drug File - Reference Terminology) [Relation: isa]
Addison's disease (Metathesaurus Names) [Relation:]
Hypoadosteronism (National Drug File - Reference Terminology) [Relation: isa]
Other adrenal hypofunction NOS (Metathesaurus Names) [Relation:]
Adrenoleukodystrophy (National Drug File - Reference Terminology) [Relation: isa]

Narrower Concepts:

Addison's disease (National Drug File - Reference Terminology) [Relation: isa]
Addison's disease (Metathesaurus Names) [Relation:]
Hypoadosteronism (National Drug File - Reference Terminology) [Relation: isa]

Adrenal Gland Diseases (National Drug File - Reference Terminology) [Relation: inverse isa]
Adrenal Gland Diseases (Metathesaurus Names) [Relation:]
Endocrine Diseases (Metathesaurus Names) [Relation:]
Endocrine Diseases (WHO Adverse Reaction Terms) [Relation:]
Adrenal cortical hypofunction (SNOMED Clinical Terms)

Relations

Concept Report Display All (continued)

Co-occurrence
data

The screenshot displays a list of MeSH terms under the heading "Associated Expressions: None found." and "Co-occurring MeSH Terms:". The terms are listed in two columns, with the right column being a zoomed-in view of the first six terms from the left column. The terms are:

- [64] Corticotropin
- [57] Hydrocortisone
- [38] Glucocorticoids
- [31] Esophageal achalasia
- [30] Dog Diseases
- [28] Adrenal Glands
- [27] Anti-Inflammatory Agents
- [26] DNA-Binding Proteins
- [25] Repressor Proteins
- [25] TRANSCRIPTION FACTORS
- [24] Retinoic Acid Receptors
- [23] Pituitary, Adrenal
- [21] Lacrimal Apparatus
- [20] Hypothalamic Hypophysial Region
- [19] Postoperative Complications
- [18] Adrenal Gland Neoplasms

Metathesaurus Advanced Search

UMLS Knowledge Source Server (UMLS SKS)
UMLS SKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

Metathesaurus Semantic Network SPECIALIST London Logout

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- ▶ [Semantic Network Resources](#)
- ▶ [Metathesaurus Resources](#)

Metathesaurus Advanced Search Options

Perform Focused Search

Focused searching allows users to search for a concept that contains either a user entered term name or a concept unique identifier (CUI). The advanced searching option allows user to restrict the results to a set of source vocabularies and may specify the method of matching the entered term name. When searching for a term name, the user may also specify the criteria to be used in matching the entered string to the UMLS contents.

Perform XML Query

The XML query facility allows users to specify a query using the eXtensible Markup Language (XML) dialect defined for the UMLS SKS. User's of this searching feature must be familiar with the internal database structures and how the data are related to each other.

Request Relational Records

The data tables used to populate the backend Oracle database can be returned to the user that match a user's input term or concept unique identifier.

- Focused Search

- Raw relational records from selected tables (files)

Metathesaurus Focused Search

The screenshot shows the UMLS Knowledge Source Server (UMLSKS) interface. The header includes the UMLSKS logo, version information (UMLSKS Version 4.2.2, UMLS Releases: 2002, 2002AB, 2002AC, 2002AD, 2003AA, 2003AB, 2003AC, 2004AA, 2004AB), and navigation links for Metathesaurus, Semantic Network, and SPECIALIST Lexicon. A 'Logout' link is also present.

The main content area is titled 'Metathesaurus Focused Search:' and contains the following steps:

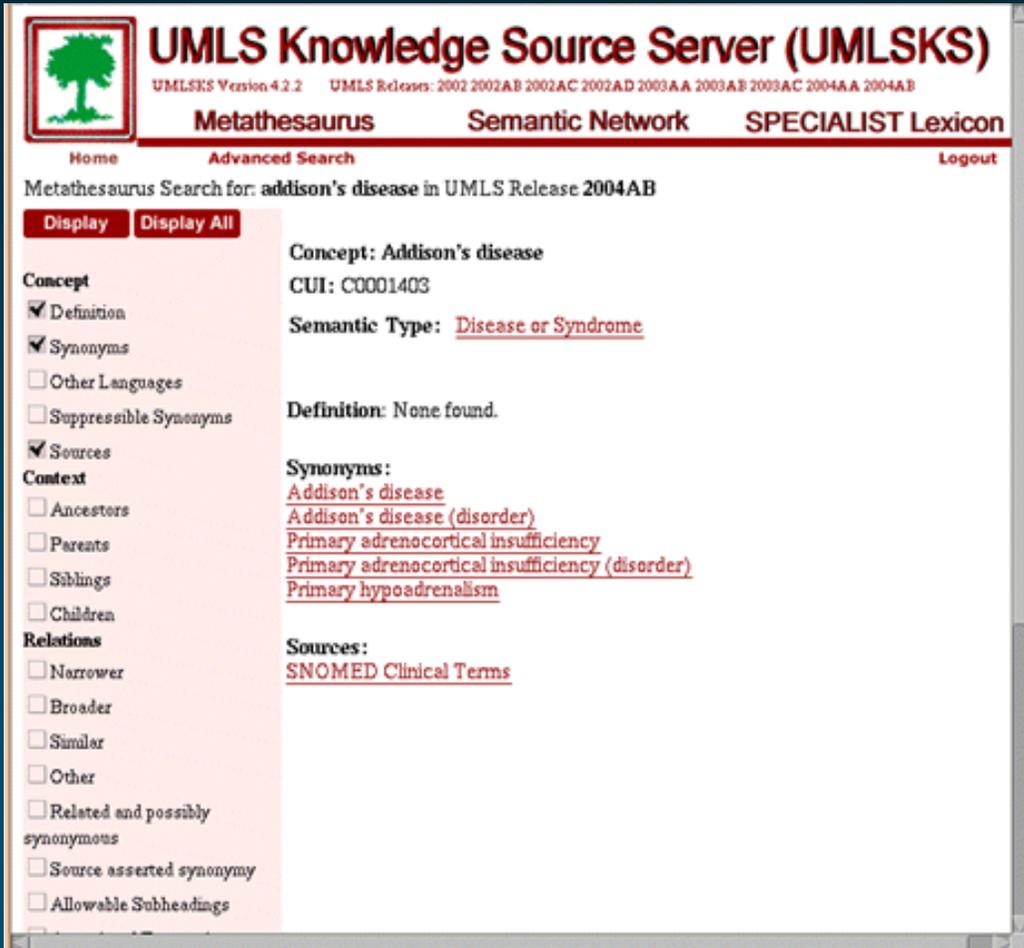
- 1) Select UMLS Release: 2004AB
- 2) Enter a term or a concept unique identifier (CUI): I
- 3) Restrict source vocabulary to:
 - All Source Vocabularies
 - Restrict to selected sources:
- 4) String Matching Criteria: Normalized string index (with a checkbox for 'Check if you want results ordered by Semantic Group')
- 5) Language: English

At the bottom, there are two buttons: 'Perform Concept Search' and 'Perform Term Search'. A list of source vocabularies is visible in a scrollable area, including ALPHEUM, Alcohol and Other Drug Thesaurus, Alternative Billing Concepts, Beth Israel Problem List, COSTAR, and CPT.

At the very bottom, a note states: 'Focused searching allows users to search for a concept that contains either a user entered term name or a concept unique identifier (CUI). The term name/CUI (e.g. C0001175) is

- UMLS Release
- Search Term
- Source Vocabularies
- String Criteria
 - Exact Match
 - Normalized string & word
 - Word
 - Truncation (left/right)
 - Approximate Match
- Language

Focused Search Concept Report



The screenshot displays the UMLS Knowledge Source Server (UMLSKS) interface. At the top, it shows the logo and version information (UMLSKS Version 4.2.2, UMLS Releases: 2002, 2002AB, 2002AC, 2002AD, 2003AA, 2003AB, 2003AC, 2004AA, 2004AB). The main navigation bar includes "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". The search results are for "addison's disease" in UMLS Release 2004AB. The interface is divided into a left sidebar with filters and a main content area. The filters include "Concept", "Context", and "Relations". The main content area displays the following information:

- Concept:** Addison's disease
- CUI:** C0001403
- Semantic Type:** [Disease or Syndrome](#)
- Definition:** None found.
- Synonyms:**
 - [Addison's disease](#)
 - [Addison's disease \(disorder\)](#)
 - [Primary adrenocortical insufficiency](#)
 - [Primary adrenocortical insufficiency \(disorder\)](#)
 - [Primary hypoadrenalism](#)
- Sources:**
 - [SNOMED Clinical Terms](#)

- UMLS Release:
2004AB
- Search Term:
addison's disease
- Source Vocabulary:
SNOMED CT
- String Criteria:
Normalized string
- Language:
English

Metathesaurus Search Relational Record

UMLS Knowledge Source Server (UMLSKS)
UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

Metathesaurus Semantic Network SPECIALIST Lexicon

Metathesaurus Relational Records Request:

Select UMLS Release:

1) Enter a term or concept unique identifier (CUI):

2) Select the UMLS table whose raw records are to be returned.

**Per
Con
Se**

Raw record return allows users to search for a concept using either a user entered concept unique identifier (CUI) or a term name which is entered into the text entry box below. Any of the UMLS tables listed in the drop down menu may be selected for return.

Pressing the *Perform Concept Search* button will update the display to show the raw records from the selected table based on the concept information found.

● UMLS Release

● Search Term

● UMLS Relational Table (Selected files)

Relational Records MRCNSO.RRF

```
CUI|LAT|TS|LUI|STT|SUI|ISPREF|AUI|SAUI|SCUI|SDUI|SAB|TTY|CODE|STR|SUPPRESS|CVF|
C0001403|CZE|P|L3180742|PF|S3708232|Y|A3910108|||D000224|MSHCZE|MH|D000224|ADDISONOVA NEMOC|
C0001403|DUT|P|L2048638|PF|S2386860|N|A6566810|||10001130|MDRDUT|LT|10001130|Addison, ziekte
C0001403|DUT|P|L2048638|PF|S2386860|Y|A3931189|||D000224|MSHDUT|MH|D000224|Addison, ziekte v
C0001403|DUT|S|L2048637|PF|S2386859|Y|A3931188|||D000224|MSHDUT|SY|D000224|Addison, syndroom
C0001403|DUT|S|L2528364|PF|S2985131|Y|A3966882|||D000224|MSHDUT|SY|D000224|Ziekte van Addisc
C0001403|DUT|S|L3205108|PF|S3732602|N|A5146733|E27.1||ICD10DUT|PT|E27.1|Primaire bijniersch
C0001403|DUT|S|L3205108|PF|S3732602|Y|A3970882|||D000224|MSHDUT|SY|D000224|Primaire bijniers
C0001403|DUT|S|L4999233|PF|S5686738|Y|A6627443|||10036696|MDRDUT|LT|10036696|primair hypoadr
C0001403|DUT|S|L4999270|PF|S5686775|N|A6627493|||10052381|MDRDUT|LT|10052381|primaire bijnie
C0001403|DUT|S|L4999270|PF|S5686775|Y|A6627494|||10052381|MDRDUT|PT|10052381|primaire bijnie
C0001403|DUT|S|L5012413|PF|S5699917|Y|A6645695|||10013096|MDRDUT|LT|10013096|ziekte van Addi
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388276|||A0D|DE|0000006012|Addison's disease|0||
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388277|||0060-3321|CSP|PT|0060-3321|Addison's diseas
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388279|||LCH|PT|U000061|Addison's disease|0||
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388280|||10001390|MDR|LT|10001130|Addison's disease|
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388281|||RCD|PT|C1541|Addison's disease|3||
```

SPECIALIST Lexicon Searching

- Select Tab along top
- Quick search

Netscape: UMLS Knowledge Source Server (UMLSKS)

File Edit View Go Communicator Help

Back Forward Reload Home Search Netscape Print Security Shop Stop

Bookmarks Location: <http://umlsks.nlm.nih.gov/kss/servlet/Turbine/act:> What's Related

Members WebMail Connections BizJournal SmartUpdate Mktplace

 **UMLS Knowledge Source Server (UMLSKS)**
UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

Metathesaurus Semantic Network SPECIALIST Lexicon

Logout

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- ▶ [Semantic Network Resources](#)
- ▶ [Metathesaurus Resources](#)

Quick Search

Select UMLS Release:

Enter search value:

Metathesaurus Concept Search **Semantic Network Search** **SPECIALIST Lexicon Search**

[Search Tips...](#) [Search Tips...](#) [Search Tips...](#)

Advanced Searches

[Metathesaurus Advanced Search](#)

Facilitates advanced searching of the UMLS Metathesaurus, including restricting vocabularies, performing batch searches, performing XML queries, and using a command-line type interface.

What's New

- ▶ 2004 AB Metathesaurus now available to download and searching for those that have signed the new license agreement!
- ▶ UMLSKS Version 4.3 released on August 30, 2004 for 2004 AB download access and searching.

Semantic Network Browser

Allows browsing of the hierarchies for the Semantic Network.

100%

SPECIALIST Lexicon Search



UMLS Knowledge Source Server (UMLS SKS)

UMLS SKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

[Metathesaurus](#) [Semantic Network](#) [SPECIALIST Lexicon](#)

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- [Developer's API Javadocs](#)

SPECIALIST Lexicon

The [SPECIALIST Lexicon](#) is an English language lexicon containing many biomedical terms. The lexicon entry for each word or term records syntactic, morphological, and orthographic information.

Lexical entries may be single or multi-word terms.

View Lexical Records for:

SPECIALIST Lexical Record



UMLS Knowledge Source Server (UMLSKS)

UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

[Metathesaurus](#)

[Semantic Network](#)

[SPECIALIST Lexicon](#)

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Specialist Lexical Record

```
|base=Addison's disease  
entry=E0000160  
cat=noun  
variants=uncount  
variants=reg
```

View "[Addison's disease](#)" in relational format.

Semantic Network Searching

- Select Tab along top
- Quick search
- Advanced Search on right-hand side

Netscape: UMLS Knowledge Source Server (UMLSKS)

File Edit View Go Communicator Help

Back Forward Reload Home Search Netscape Print Security Shop Stop

Bookmarks Location: <http://umlsks.nlm.nih.gov/kss/servlet/Turbine/act:> What's Related

Members WebMail Connections BizJournal SmartUpdate Mktplace

 **UMLS Knowledge Source Server (UMLSKS)**
UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

Metathesaurus Semantic Network SPECIALIST Lexicon

Logout

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- ▶ [Semantic Network Resources](#)
- ▶ [Metathesaurus Resources](#)

Quick Search

Select UMLS Release:

Enter search value:

Metathesaurus Concept Search **Semantic Network Search** **SPECIALIST Lexicon Search**

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Metathesaurus Advanced Search

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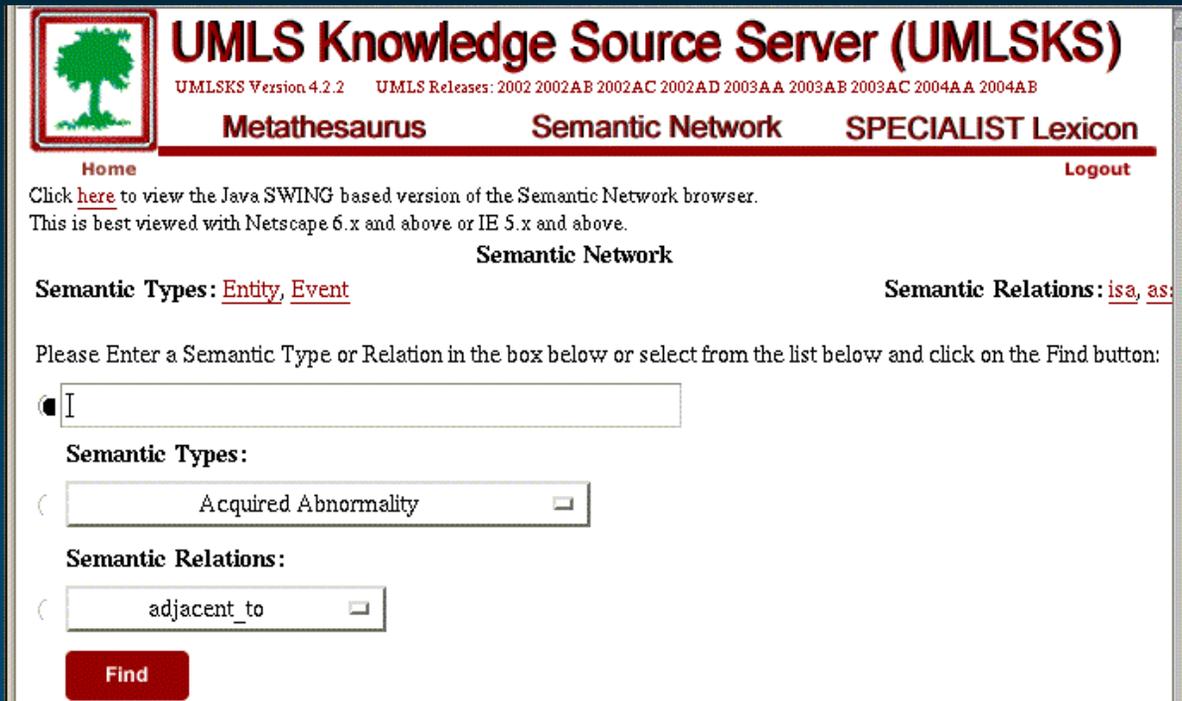
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Semantic Network Browser

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100%

Semantic Network Search



UMLS Knowledge Source Server (UMLSKS)
UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

[Home](#) [Semantic Network](#) [SPECIALIST Lexicon](#) [Logout](#)

Click [here](#) to view the Java SWING based version of the Semantic Network browser.
This is best viewed with Netscape 6.x and above or IE 5.x and above.

Semantic Network

Semantic Types: [Entity](#), [Event](#) Semantic Relations: [isa](#), [as](#)

Please Enter a Semantic Type or Relation in the box below or select from the list below and click on the Find button:

Semantic Types:

Semantic Relations:

- Enter search string
- OR-
- Select semantic type
- OR-
- Select semantic relation

Semantic Relationship Validator

Semantic Type Clinical Drug

The screenshot shows the 'Metathesaurus Semantic Network' interface. At the top, there are navigation links: Home, Metathesaurus, Semantic Network, and SPECIALIST Lexicon. Below this, there is a search area with a text input field and a 'Find' button. To the right of the search area, there are three radio buttons: 'Meta Concepts', 'Relations', and 'Raw Records'. The main content area displays the 'Semantic Type: Clinical Drug' with its TUI (T200) and a definition: 'A pharmaceutical preparation as produced by the manufacturer. The name usually includes the substance, its strength, and the form, but may include the substance and only one of the other two items.' Below the definition, there is a hierarchical tree of semantic types, starting with 'Entity' and branching into 'Physical Object', 'Organism', 'Plant', 'Alga', 'Fungus', 'Virus', 'Rickettsia or Chlamydia', 'Bacterium', 'Animal', 'Invertebrate', 'Vertebrate', 'Amphibian', 'Bird', 'Fish', 'Reptile', and 'Mammal'.

- Browse ST hierarchy
- View Concepts with ST
- View Relations valid for the ST
- View Raw Relational Records

UMLSKS Downloads

Downloads

- ▶ [UMLS Knowledge Sources](#)
- ▶ [RxNorm Files](#)
- ▶ [SNOMED CT®](#)
- ▶ [Mappings](#)
- ▶ [VA/KP Problem List](#)
- ▶ [Developer's API](#)

UMLS Release Files

SNOMED CT files in Native format

SNOMED CT ®

Download SNOMED CT international releases, English and Spanish, from this page.

July 2007 International Release of SNOMED CT files

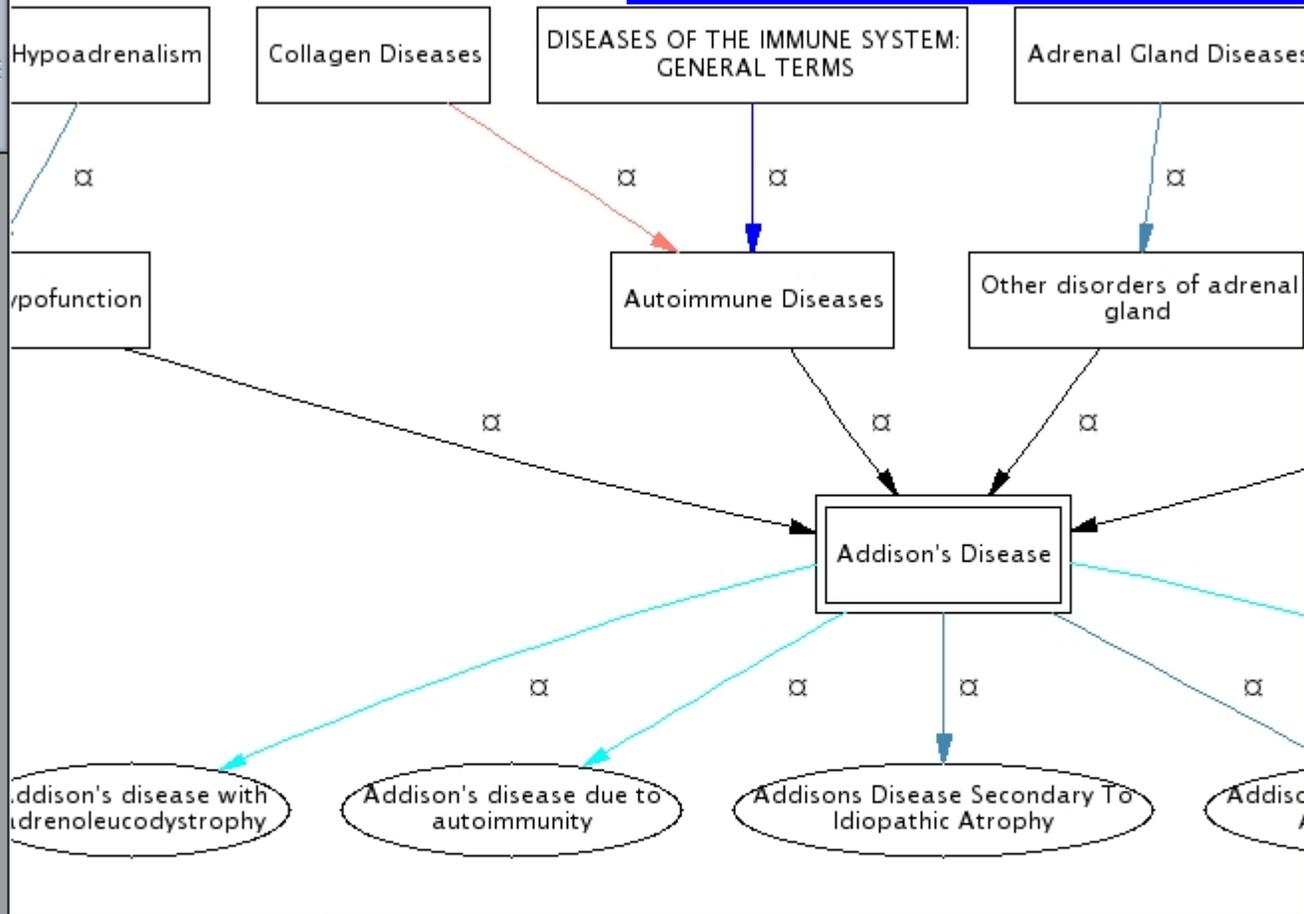
File Name	Description
SNOMED CT Essential 20070731.zip (127M)	Contains the International Release Essential Resources including SNOMED CT terminology, cross maps, subsets, documentation, and other resources
SNOMED CT Optional 20070731.zip (2.6M)	Contains the International Release Option Resources including the ICD-9-CM cross map, and US drug extensions
SNOMED CT User Guide.pdf (507K)	Explains the capabilities, content, principles used to model the terminology, and uses from a content perspective
SNOMED CT Technical Implementation Guide.pdf (1.0M)	Guidelines and advice about the design of applications using SNOMED CT, covers terminology services, entering and storing information, and migration of legacy information
SNOMED CT Technical Reference Guide.pdf (2.9M)	Reference material for the current release of SNOMED CT including file layouts, field sizes, required values and their meanings, and high level data diagrams.

SNOMED CT (Spanish) April 2007

[SNOMEDCT Spanish 2007 04 20.zip](#) | [SNOMED CT native files in Spanish](#)

Semantic Navigator

- Siblings**
- Concepts & Ideas**
- Clinical Syndromes
- Disorders**
- Acquired Immunodeficiency Syndrome
 - Acute adrenal insufficiency
 - Addisonian crisis
 - Adrenal atrophy
 - Adrenal calcification
 - Adrenal hemorrhage
 - Adrenal infarction
 - Adrenal insufficiency due to adrenal metastasis
 - Adrenogenital Syndrome
 - Allergic arthritis
 - Angelman Syndrome
 - Asperger syndrome
 - Autoerythrocyte sensitivity



- Other Related Concepts**
- Disorders**
- Addisonian crisis
 - Autoimmune Syndrome Type II, Polyglandular
 - Tuberculosis
 - Tuberculosis of adrenal glands
 - Tuberculous Addison's disease
- (5 other related)
- Co-occurring Concepts**
- Anatomy**
- Adrenal Cortex [14]
 - Adrenal Glands [17]
 - Liver [2]
 - Tears body substance [2]
 - X Chromosome [3]
- Chemicals & Drugs**

BCI Addison's Disease **LEGEND ***

Start again Apply new parameters

Restrict to vocabulary: Show all

Highlight vocabulary: Nothing

UMLS data: UMLS_2002

Type of hierarchical: All Parent/Child only

Similar Concepts

- Adrenal cortical hypofunction

(1 concept)

Closest MeSH Terms

Main Headings

- Addison's Disease

Links to UMLS Resources

- NLP & Lexical Resources
 - MetaMap Transfer (MMTx)
 - Word Sense Disambiguation (WSD) Test Collection
- Semantic Network
 - Semantic Navigator
 - Semantic Groups
- Metathesaurus
 - String Properties

Resources

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- ▶ [Semantic Network Resources](#)
- ▶ [Metathesaurus Resources](#)

Questions?

Summary

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 - Metathesaurus
 - Semantic Network
 - SPECIALIST Lexicon and Lexical Tools
- UMLSKS
 - Remote access and browser
 - Download files
 - Documentation
- MetamorphoSys
 - Installs UMLS, customizes Metathesaurus
 - RRF Browser

Bibliography of UMLS Articles

- Search MEDLINE[®]/PubMed[®] for recent research journal articles on the UMLS (1997 to present)
- Comprehensive Bibliography 1986-1996
 - Selden, Catherine
Unified Medical Language System (UMLS): January 1986 through December 1996 : 280 citations / prepared by Catherine R. Selden, Betsy L. Humphreys. -- Bethesda, Md. (8600 Rockville Pike, Bethesda 20894) : U.S. Dept. of Health and Human Services, Public Health Service, National Institutes of Health, National Library of Medicine, Reference Section ; Pittsburgh, PA : Sold by the Supt. of Docs., U.S. G.P.O., 1997.
-- (Current bibliographies in medicine ; 96-8)

References

- UMLS as a research project
 - Lindberg, D. A., Humphreys, B. L., & McCray, A. T. (1993). The Unified Medical Language System. *Methods Inf Med*, 32(4), 281-91.
 - Humphreys, B. L., Lindberg, D. A., Schoolman, H. M., & Barnett, G. O. (1998). The Unified Medical Language System: an informatics research collaboration. *J Am Med Inform Assoc*, 5(1), 1-11.
- Short presentation
 - Bodenreider, O. (2004) The Unified Medical Language System (UMLS): integrating biomedical terminology. *Nucleic Acids Res*, 32(Database issue), D267-70.

UMLS Documentation and Support

- UMLS homepage
 - <http://umlsinfo.nlm.nih.gov/>
- UMLSKS homepage
 - <http://umlsks.nlm.nih.gov>
- UMLSUSERS-L
 - subscribe to discussion list
- NLM Customer Service email:
 - custserv@nlm.nih.gov

Explore

- Register: sign the license agreement
- Create UMLSKS account
- Explore Knowledge Sources
- Download files or request DVD
- Create subsets using MetamorphoSys



Center
Drive

Library
of
Medicine

Thank you